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(54) Title: CHARATERIZATION OF SINGLE NUCLEOTIDE POLYMORPHISMS IN CODING REGIONS OF HUMAN GENES			
(57) Abstract			
<p>The invention provides nucleic acid segments of the human genome, particularly nucleic acid segments from the coding region of a gene, including polymorphic sites. Allele-specific primers and probes hybridizing to regions flanking or containing these sites are also provided. The nucleic acids, primers and probes are used in applications such as phenotype correlations, forensics, paternity testing, medicine and genetic analysis.</p>			

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CHARACTERIZATION OF SINGLE NUCLEOTIDE POLYMORPHISMS
IN CODING REGIONS OF HUMAN GENES

RELATED APPLICATION

This application claims the benefit of U.S. Provisional Application Serial
5 No. 60/127,248, filed March 31, 1999, the entire teachings of which are incorporated
herein by reference.

GOVERNMENT SUPPORT

The invention was supported, in whole or in part, by grant 5-P50-HG00098-
09 SNP from the National Institutes of Health (NCHGR) and grant 1-R01-
10 HL61774-01 from the National Institutes of Health (NHLBI). The Government has
certain rights in the invention.

BACKGROUND OF THE INVENTION

A major goal in human genetics is to understand the role of common genetic
variants in susceptibility to common diseases (N. Risch and K. Merikangas, *Science*,
15 273:1516 (1996.); *E. S. Lander, Science, 274:536 (1996); F.S. Collins, et al.,*
Science, 278:1580 (1997)). This requires assembling an extensive catalogue of
single-nucleotide polymorphisms (SNPs) and performing systematic association
studies for particular diseases.

The human population has relatively limited genetic diversity, reflecting its
20 young age and historically small size (F. J. Ayala *et. al., Proc. Natl. Acad. Sci.,*
91:6787 (1994)). Given the restricted nature of the allelic spectrum, some authors
have recently suggested that it should eventually be possible to collect all common
SNPs in the human population and have hypothesized that such common variants
may underlie much of the genetic risk of common disease (N. Risch and K.
25 Merikangas, *Science, 273:1516 (1996.); E. S. Lander, Science, 274:536 (1996); F.S.*
Collins, et al., Science, 278:1580 (1997)). This is in contrast to the situation for rare

Collins, *et al.*, *Science*, 278:1580 (1997)). This is in contrast to the situation for rare genetic diseases, which are primarily caused by a large number of distinct alleles that are recent, rare and highly penetrant. Important examples of associations to common (>1%) alleles include the ApoE4 allele in Alzheimer's disease, the Factor
5 V^{Leiden} allele in deep-venous thrombosis, and the CCR5- $\Delta 32$ in resistance to HIV infection (A. M. Saunders *et al.*, *Neurology*, 43:1467 (1993); R. M. Bertina, *Nature*, 369:64 (1994); M. Dean *et al.*, *Science*, 273:1856 (1996)). The most relevant variants are likely to be those in coding and regulatory regions of genes.

SUMMARY OF THE INVENTION

10 As described herein, the nature of SNPs in the coding regions of human genes has been explored. SNPs were identified in 106 genes relevant to cardiovascular disease, endocrinology and neuropsychiatry, by screening an average of 114 independent alleles using two independent screening methods. To ensure high accuracy, all reported SNPs were confirmed by DNA sequencing. A total of
15 545 SNPs were identified, including 395 coding-regions SNPs (cSNPs) divided roughly equally between those causing synonymous and non-synonymous changes. The cSNPs most likely to influence disease, those that alter the amino acid sequence of the encoded protein, show strikingly different properties: they occur at a lower rate and with lower allele frequencies. This likely reflects selection acting against
20 deleterious alleles during human evolution. The lower allele frequency of cSNPs has important implications for the number of chromosomes that must be sampled to construct a comprehensive catalogue of human cSNPs.

The invention relates to a gene which comprises a single nucleotide polymorphism at a specific location. In a particular embodiment the invention
25 relates to the variant allele of a gene having a single nucleotide polymorphism, which variant allele differs from a reference allele by one nucleotide at the site(s) identified in Figures 5A-5Q. Complements of these nucleic acid segments are also included. The segments can be DNA or RNA, and can be double- or single-stranded. Segments can be, for example, 5-10, 5-15, 10-20, 5-25, 10-30,
30 10-50 or 10-100 bases long. The invention further relates to gene products encoded by genes and oligonucleotides of the invention.

The invention further provides allele-specific oligonucleotides that hybridize to a gene comprising a single nucleotide polymorphism or to the complement of the gene. These oligonucleotides can be probes or primers.

The invention further provides a method of analyzing a nucleic acid from an individual. The method determines which base is present at any one of the polymorphic sites shown in Figures 5A-5QQQQQQQ. Optionally, a set of bases occupying a set of the polymorphic sites shown in Figures 5A-5QQQQQQQ is determined. This type of analysis can be performed on a number of individuals, who are tested for the presence of a disease phenotype. The presence or absence of disease phenotype is then correlated with a base or set of bases present at the polymorphic site or sites in the individuals tested.

BRIEF DESCRIPTION OF THE DRAWINGS

Figure 1 is a graph showing minor allele frequency by polymorphism type. The percentage of cSNPs having minor allele frequency classified as low (<5%), medium (5-15%) or high (>15%) frequency is displayed for synonymous, non-synonymous and non-coding SNPs.

Figure 2 is a graph showing the distribution of nucleotide diversity. Normalized frequency of variant sites, θ , was calculated for the coding region of each gene. The graph shows the percentage of genes having θ in the indicated range.

Figures 3A and 3B are a table showing a summary of polymorphisms in 106 human genes described herein. Column 1 shows the name of the gene as used in Online Mendelian Inheritance in Man. Column 2 shows the number of coding base pairs screened. Column 3 shows the number of synonymous (or silent) polymorphisms identified. Column 4 shows the number of non-synonymous polymorphisms identified. Column 5 shows the number of non-coding base pairs screened. Column 6 shows the number of non-coding polymorphisms, including those in introns and untranslated regions (UTR), identified.

Figure 4 is a table showing polymorphism rates for different classes of sites.

Figures 5A-5QQQQQQQ are a table showing the specific polymorphisms identified in the genes studied as described herein. Column 1 shows the laboratory

designation for the polymorphism. Column 2 shows the name of the gene as used in Online Mendelian Inheritance in Man. Column 3 shows the reference nucleotide which occupies the polymorphic site in the reference allele. Column 4 shows the variant nucleotide which occupies the polymorphic site in the variant allele.

- 5 Column 5 shows the reference amino acid encoded by the codon which contains the polymorphic site in the reference allele. Column 6 shows the variant or alternate amino acid encoded by the codon which contains the polymorphic site in the variant allele. Column 7 indicates whether the polymorphism is located in the coding or non-coding region of the gene. Column 8 shows the assay number in which the
10 polymorphism was assessed. Columns 9 and 10 show the forward and reverse primers, respectively, which were used to identify the polymorphism. Column 11 shows the sequence of the gene used in the assay, with the polymorphic site indicated by brackets and the primers shown in capital letters. Column 12 shows the total number of nucleotides given in Column 11.

15 DETAILED DESCRIPTION OF THE INVENTION

- There is a rich literature concerning nucleotide variation in model systems, particularly in *Drosophila* (E. N. Moriyama and J. R. Powell., *Mol. Biol. Evol.*, 13:261 (1996)), but sequence variation in human genes has been studied only in limited ways. A small number of studies have focused on individual genes (such as
20 beta-globin and lipoprotein lipase) in many individuals, and one study examined 49 genes by comparing two independent sequences deposited in public databases (R. M. Harding *et. al.*, *Am. J. Hum. Genet.*, 60:772 (1997); D. A. Nickerson *et. al.*, *Nature Genetics*, 19:233 (1998); W. H. Li and L.A. Sadler, *Genetics* 129:513 (1991)). To perform a more comprehensive survey, as described herein, a collection
25 of 106 genes were selected whose protein products play important roles in the cardiovascular, endocrine and neurological systems (Figures 3A-3B and Figures 5A-5Q). Gene sequences were obtained from the Genbank and TIGR databases. Where multiple sequence depositions were available, a consensus sequence was derived. Determination of coding sequence, untranslated regions and
30 intronic regions was based on annotation in the public database, although internal

checks were performed to ensure accurate determination of start and stop codons, open reading frames and the like.

The genes were chosen because of their relevance to common, clinically significant diseases, such as coronary artery disease, diabetes, and schizophrenia. They encode proteins involved in coagulation, lipid metabolism, energy metabolism, neuroendocrine physiology, neurotransmission and central nervous system development. Variation in these genes was studied in a sample including Caucasians, African-Americans, African Pygmies and Asians, with an average of 114 chromosomes screened for each gene. Of the samples screened, 30 were from Caucasian individuals, 14 from Asian, 10 African American and 7 Africans. The average number of individuals successfully screened for each gene was 57, with the precise number successfully screened varying among genes. Cell lines were obtained from Coriell Cell Repository, and DNA prepared according to standard protocols. In addition, 10 of the Caucasian samples used in this study were obtained as anonymous blood samples from the Physician's Health Study (gift of Charles Hennekens and J. Michael Gaziano). The sample size provides greater than 65% power to detect alleles with frequency of 1%.

Overall, the sample of 114 chromosomes was screened for SNPs in a total of 195.4 kb, consisting of 135.8 kb of coding regions and 59.6 kb from adjacent non-coding region (untranslated region (UTR) and introns). Sequences were amplified by the polymerase chain reaction (PCR) and screened by two independent methods. The first method involved hybridization of labeled PCR products to variant detector arrays (VDAs) (that is, high density DNA probe arrays containing oligonucleotides specific for the sequences under study) (M. Chee *et al.*, *Science*, 274:610 (1996); D. G. Wang *et al.*, *Science*, 280:1077 (1998)); variant sequences typically give rise to altered hybridization patterns. These chips contained variant detector arrays (VDA) (M. Chee *et al.*, *Science* 274:610 (1996)).

Using VDAs, candidate SNPs were identified using a combination of three algorithms followed by visual inspection. For each base position and strand queried there are four VDA features: one contains the expected base (the reference sequence) in the central position and the other three features contain central substitution bases (in the background of the reference sequence). The base-calling

algorithm looked for positions at which hybridization to a substitution base gives a stronger signal than the reference base. The second algorithm (mutant fraction) examined the reference base and each one of the substitution bases in turn and calculates the fraction of signal present in the non-reference base. The final
5 algorithm (footprint detection) depends upon a loss of signal at the reference positions surrounding a nucleotide substitution. These algorithms are combined to yield a confidence score of "certain" or "likely" for each candidate polymorphism. Two analysts independently scored the data, and candidate polymorphisms found by either observer were included in subsequent confirmation tests. PCR assays
10 spanning each exon were designed using Primer 3.0 release 0.7. PCR was performed according to standard protocols, and assays destined to be hybridized to the same chip design were pooled together. Chip samples were prepared and hybridized as described in D.G. Wang *et al.* (*Science* 280:1077 (1998)), except that pools consisting of about 100 assays contained 5-6 μ g of amplified material. In all,
15 854 assays (average size of 300 bp, covering 106 genes) were amplified from each individual and were hybridized to 12 distinct chip designs. The probe arrays were designed to query only the coding sequence for some genes, while other genes contained the entire mRNA and/or surrounding intron (Figures 3A-3B). The second method involved subjecting PCR products to Denaturing HPLC (dHPLC) (P. J.
20 Oefner and P. A. Underhill, *Am. J. Hum. Genet.*, 57:A266 (1995)) at a critical temperature; heterozygous individuals typically give rise to heteroduplex products with altered denaturation and migration properties.

Sequences were amplified as above except that the final extension in the PCR protocol was followed by denaturation and slow reannealing to allow
25 heteroduplex formation. A total of 6 μ l of each individual PCR product was injected into Wave DNA Fragment Analysis System (Transgenomic). A total of 592 of the VDA assays (covering the 89 genes attempted with this method) were successfully screened by DHPLC. Only assays of >160 base pairs were used for DHPLC, because shorter assays performed unreliably for mutation detection. The
30 DHPLC parameters (percentage of acetonitrile, column temperature) used for each fragment were automatically calculated using a novel predictive algorithm, and DHPLC traces were analyzed using the clustering program ASH v2.0. A scoring

algorithm was developed based upon the similarity score by ASHv2.0 and contour of the elution profile.

Because both screening methods can generate to a significant number of false positives, it was important to confirm every reported SNP. Samples implicated
5 by either method as containing a candidate SNP were thus subjected to fluorescent dideoxy sequencing, either to confirm the presence of the SNP (in the case of the chip) or to identify and confirm the presence of the SNP (in the case of DHPLC). Such confirmation proved essential for eliminating false positives.

Candidate SNPs were either validated (if found by VDAs) or identified (if
10 implicated by DHPLC) by DNA sequencing. For this purpose, sequences were amplified with PCR primers tailed with standard M13 sequencing sites (-21 forward and -28 reverse) and conventional dye-primer sequencing was performed on ABI 377 sequencers. For candidate SNPs discovered by VDAs, one individual was chosen (a candidate homozygous variant, when available, or a candidate
15 heterozygote) and sequencing was performed on one strand to confirm by visual inspection the presence of the SNP at the indicated position. For amplicons found to be polymorphic by DHPLC, two individuals were selected representing each distinct elution pattern observed and were sequenced on both strands to discover the variant base or bases. Sequences were base-called by the Phred program, assembled
20 by the Phrap program, and polymorphism candidates were identified by the PolyPhred program (D. A. Nickerson *et. al.*, *NAR*, 25:2745 (1997)). All results were visually inspected by at least two observers.

The overall false positive rate for VDAs was 45%. The rate was much lower (about 10%) for certain chip designs, synthesis protocols, and for candidate
25 polymorphisms scored as "certain." The false positive rate among fragments displaying an altered elution pattern by DHPLC was similar (40%). The false positive rates reflect the thresholds employed for declaring a candidate SNP, which were chosen to ensure high sensitivity.

A total of 545 SNPs were identified in the 195 kb surveyed, consisting of
30 150 non-coding SNPs and 395 cSNPs. Results from these studies are shown in the Figures. The complete data are available on the web site
http://www.genome.wi.mit.edu/cvar_snps; access to this website can be gained

using the guestname "snp_pilot" and the password "noyne". In the future, access to this website may be available to the public, and thus, no guestname or password may be needed.

To directly determine the false-negative rate of the screen, conventional
 5 DNA sequencing was performed on ten of the genes (THPO, TBAX2R, PTHLH, IGF2, HTR2A, HTR1A, GHR, GABRB1, F10, and CYP11B1) spanning 25.2 kb in twenty individuals. Sequencing was performed on both strands using dye-primer chemistry and sequence traces were interpreted using PolyPhred (D.A. Nickerson *et al.*, *NAR*, 25:2745 (1997)). VDA analysis identified 85% of variants found by direct
 10 sequencing, while DHPLC identified 87% of the variants found by direct sequencing. In regions screened by both VDAs and DHPLC, the combination of the two methods identified 100% of the polymorphisms found by direct sequencing.

Overall, about one-third of individuals were screened with both methods, and one-third were screened with each of the two methods alone. (For some genes, the
 15 non-coding regions were screened only by DHPLC.) It is estimated that the false negative rate over the entire study to be about 15% for regions screened by one method, and negligible for sequences screened by both methods. The total number of true polymorphisms not identified is estimated to be less than 10%.

A SNP survey can be characterized in terms of either K, the observed
 20 number of variant sites, or p, the observed heterozygosity per bp. Because K increases with the number of chromosomes (n) studied and the total sequence length L, it is preferable to use the normalized number of variant sites

$$\hat{\theta} = K / \left(\sum_{i=1}^{n-1} i^{-1} \right) L \text{ which corrects for sample size. Under the neutral}$$

theory of molecular evolution and infinite sites model, θ and π are both estimators
 25 of the population genetic parameter $\theta = 4N\mu$ (Li, *Molecular Evolution*, Sinauer Associates (1997), Canada).

SNPs were found at a similar overall frequency in coding and non-coding regions. SNPs in coding region occurred at a frequency of 1 per 344 bp, corresponding to $\hat{\theta} = 5.47 \times 10^{-4}$ and $\pi = 5.07 \times 10^{-4}$. Interestingly, SNPs were
 30 observed in non-coding DNA at a similar frequency of 1 per 397 bp. The

normalized number of variant sites was $\theta = 4.93 \times 10^{-4}$, and the mean heterozygosity (π) = 5.05×10^{-4} (Figure 4). Calculations of π involve allele frequencies. Polymorphisms identified by DHPLC alone were excluded because we did not sequence all of the samples showing a variant DHPLC pattern and thus could not be certain of allele frequency. The estimates of π were thus based on 411 of 545 polymorphisms. Although the VDAs were designed for polymorphism discovery rather than genotyping, the estimated allele frequencies proved to be quite accurate. Specifically, genotyping assays (employing single-base extension assays) for 25 SNPs yielded allele frequencies that differed by an average of only 2% from those estimated on the basis of genotypes inferred from the VDA. For both classes, the similar values for θ and π is consistent with a population evolving according to neutral expectations.

The 395 cSNPs were roughly equally divided between synonymous (203 cSNPs) and non-synonymous (192 cSNPs) changes. Since approximately two-thirds of random mutations would alter an amino acid, the fact that non-synonymous cSNPs comprise slightly less than half of the cSNPs implies strong selection against amino-acid altering changes. To address this issue more directly, the nucleotide diversity was examined at four-fold degenerate sites, two-fold degenerate sites, and non-degenerate sites. Changes at four-fold degenerate sites produce only synonymous changes, while those at non-degenerate sites are always non-synonymous. Nucleotide diversity (θ) was 9.64×10^{-4} at four-fold degenerate sites, 6.85×10^{-4} at two-fold degenerate sites, and 3.70×10^{-4} at non-degenerate sites. Assuming that mutations occur at an equal rate at both classes of sites, non-synonymous variants survive to be detected in such a survey at only 38% of the rate of synonymous changes.

The force of selection is also evident in comparing non-synonymous cSNPs causing a non-conservative amino acid alteration with those causing a conservative amino-acid change. Conservative and non-conservative amino acid substitutions were defined for this analysis according to the BLOSUM62 matrix, used in sequence comparison (S. Henikoff and J. G. Henikoff, *PNAS*, 89:10915 (1992)). Conservative changes were those having a positive or neutral sign in the matrix, while non-conservative changes were those having a negative value. Non-conservative

cSNPs represent only 36% of the non-synonymous cSNPs, whereas randomly distributed mutations would be expected to produce a higher proportion (52%) of non-conservative changes. The proportion of non-synonymous SNPs expected to cause a non-conservative amino acid substitution was determined based on the actual codon usage in the 106 genes studied, the known frequencies of transitions and transversions, and the definition of non-conservative changes employed in the BLOSUM62 matrix. This implies that non-conservative cSNPs survive to be detected in such a survey at only about half of the rate of conservative, non-synonymous cSNPs.

10 The various types of SNPs differ not only in the rate of their occurrence, but also in the frequency of their minor alleles. This can be seen in several ways. When SNPs are classified according to whether the frequency of the minor allele was high ($\geq 15\%$), intermediate (5-15%) or low ($\leq 5\%$), it is clear that the non-synonymous cSNPs were enriched in low frequency alleles compared to the rest of the collection (Figure 1). The distribution of non-synonymous allele frequencies was significantly different than that of synonymous changes ($p=0.02$, Kolmogorov-Smirnov test). Indeed, more than half (58%) of non-synonymous cSNPs were found at a frequency below 5%, with this effect evident for both conservative and non-conservative substitutions.

20 The effect of selection can also be inferred by considering the average frequency of the minor allele: it is 8% for non-conservative cSNPs, 11% for conservative but non-synonymous cSNPs, and 14% for both synonymous cSNPs and non-coding SNPs. In addition, the lower allele frequency of non-synonymous cSNPs is reflected in the fact that the heterozygosity π is lower than the normalized rate of variant sites θ for this class of SNPs (Figure 4). This divergence is in the direction predicted by the action of purifying selection, although it falls short of statistical significance. Tajima's D was non-significant. (F. Tajima, *Genetics*, 123:545 (1989).

30 The distribution of SNPs among the 106 genes was explored, with an eye toward detecting differential effects of selection among genes. The number of cSNPs per gene ranged from 37 for Factor V to 0 for thirteen of the genes, and the normalized rate, θ , similarly showed considerable variation (Figure 2). The

observed variation in nucleotide diversity is similar in magnitude to that observed for *Drosophila* (E. N. Moriyama and J. R. Powell., *Mol. Biol. Evol.*, 13:261 (1996)). Variation among genes could be due to many factors (D. J. Begun and C. F. Aquadro, *Nature*, 356:519 (1993); Nachman *et. al.*, *Genetics*, 150:1133 (1998)).

- 5 The fact that non-synonymous cSNPs show a somewhat wider variation than synonymous cSNPs (the coefficient of variation is 20% larger for the former class) is consistent with differences in selective constraints among loci, but the difference falls well below statistical significance. A variety of population genetic tests are available for testing selection at individual loci (M. L. Wayne and K. L. Simonson, 10 *Trends and Ecology and Evolution*, 13:236 (1998)).

The age of a SNP allele has important implications for its use in human genetic studies. Recently-occurring SNP alleles are more likely to show extensive linkage disequilibrium (retention of the ancestral haplotype on which they arose) as compared to older SNPs. Such linkage disequilibrium can provide a powerful tool 15 in identifying disease genes (E. S. Lander, N.J. Schork, *Science*, 265:2037 (1994)). Although the precise age of the SNPs could not be assessed from these studies, characterization of which allele preceded human speciation and which arose thereafter was sought. To determine the ancestral human allele, each corresponding gene was sequenced from the common chimpanzee (*P. troglodytes*). Each assay 20 used in the human survey was amplified from a single chimpanzee (DNA gift of Kristin Ardlie) and subjected to dye-primer sequencing on both strands. A single chimpanzee sample will accurately reveal the ancestral allele except in cases where the site has mutated and fixed during the chimpanzee evolution or is polymorphic in the chimpanzee population and happened to be homozygous for the non-ancestral 25 allele. These two cases are quite rare (probably less than 2%) and thus have been neglected for the purpose of estimating overall rates. A human allele was considered to be ancestral if it was present in the homozygous state in the chimpanzee sample. A total of 136 kb of chimpanzee sequence was obtained, revealing an inter-species divergence of 0.6% in the regions studied.

- 30 An elegant result in theoretical population genetics predicts that the probability that a neutral allele represents the ancestral state should be equal to its frequency in the population (G. A. Watterson and H. A. Guess, *Theoretical*

Population Biology, 11:141 (1977)). The minor allele should thus represent the ancestral state in a predictable proportion of cases. The ancestral allele and minor allele frequency was determined for 267 of the reported SNPs. For 3 of the 267 SNPs, the chimpanzee was homozygous for a third allele differing from both of the
5 current human alleles. This is consistent with the overall 0.6% nucleotide sequence divergences seen between human and chimpanzee. Among polymorphisms with a minor allele frequency below 10%, the average allele frequency was 3% and the proportion that was ancestral was 7% (11/158) of cases. Among polymorphisms with minor alleles exceeding 10%, the mean frequency was 28% and the proportion
10 that were ancestral was 32% (35/109). These results thus agree remarkably well with the theoretical prediction, providing the first reported test of this prediction in humans. It therefore follows that the minor SNP allele need not be the younger allele; this has implications for linkage disequilibrium mapping.

The distribution of SNPs among Caucasian, African-American, African and
15 Asian samples was also examined. Although the vast majority of SNPs were seen in multiple groups, there was a statistically significant excess of SNPs that were seen in only one of the sub-groups. The probability that a SNP occurring $k > 1$ times in an overall sample of n individuals would be found entirely within a given subset of m individuals is $B(n,k)/B(m,k)$, where $B(x,y)$ is the binomial coefficient $x!/(x-y)!y!$. In
20 this fashion, the probability that each individual SNP would be confined to a particular ethnic subgroup within the sample was calculated and these probabilities were summed to obtain the number of SNPs expected to be confined to the group within the sample. The fact that a SNP is found only within one group in the sample does not necessarily imply that it is private to that group within the general
25 population, owing to the small sample size, but it can be used as an indication of substructure. The number of SNPs with $k > 1$ confined to the, African-Americans, African Pygmies, Caucasians, and Asians was 17, 17, 12, and 9, as compared to expectations of 3.02, 1.34, 8.62, and 1.81. Not surprisingly, the greatest excess was seen for SNPs found in the African-American and African samples. The presence of
30 population substructure implies that construction of a comprehensive SNP database should employ a diverse set of DNA samples.

The results of this survey provide a fundamental description of sequence variation in the coding regions of human genes. These data indicate that two copies of a gene chosen from the human population will differ by roughly one base in 2 kb, corresponding to somewhat less than one heterozygous base within the coding
5 region of a typical gene. In general, there are only a handful of such cSNPs per gene that exhibit allele frequencies of at least a few percent. Accounting for both the different rate and frequency of non-synonymous SNPs, only about 40% of these observed changes will alter the encoded amino acid. The action of purifying selection during human evolution is evident from the comparatively lower rate of
10 non-synonymous cSNPs, and especially of those that create a non-conservative change. It is clear that non-synonymous cSNPs not only occur less often, but also have lower minor allele frequencies: 60% of non-synonymous cSNPs, the class likely to have the most dramatic effects on proteins, display a minor allele frequency below 5%.

15 The relative rarity of cSNPs has important implications for efforts to produce large catalogues of human variants. It has been proposed that most human SNPs could be found by performing shotgun sequencing on a handful of individuals (J. L. Weber and E. W. Myers, *Genome Research*, 7:401 (1997); J. C. Venter *et. al.*, *Science*, 280:1540 (1998)). Although such a project will surely identify many SNPs,
20 results described herein suggest that the small sample size will likely fail to identify the vast majority of cSNPs likely to have the most important biological consequences, owing to their lower average allele frequencies. A comprehensive collection of the common, non-conservative cSNPs may require surveying 50-100 chromosomes. Because coding sequence represents only about 3% of the genome, it
25 may prove inefficient to obtain such deep coverage of cSNPs by shotgun sequencing of genomic DNA. Instead, it may be more efficient to perform shotgun sequencing on cDNA libraries from multiple individuals or to amplify genes from multiple individuals, as done here.

Interestingly, a similar rate of polymorphism in coding and non-coding DNA
30 was found. Furthermore, the observed rate of nucleotide diversity at four-fold degenerate sites was nearly twice that in adjacent non-coding regions, and over twice that at non-degenerate sites (Figure 4). Similar results have been reported for

Drosophila (E. N. Moriyama and J. R. Powell., *Mol. Biol. Evol.*, 13:261 (1996)) and for a smaller human data set by Li and Sadler (R. M. Harding *et. al.*, *Am. J. Hum. Genet.*, 60:772 (1997); D. A. Nickerson *et. al.*, *Nature Genetics*, 19:233 (1998); W. H. Li and L.A. Sadler, *Genetics* 129:513 (1991)), who observed over three times
5 the nucleotide diversity at four-fold degenerate sites ($\theta = 11 \times 10^{-4}$), as compared to that in both untranslated regions and non-degenerate sites ($\theta = 3 \times 10^{-4}$). These observations suggest that non-coding DNA adjacent to coding regions may be functionally constrained to a surprising degree.

SNPs can be used to search for genes underlying complex traits in two
10 distinct ways: linkage disequilibrium (LD) studies and association studies (E. S. Lander, N.J. Schork, *Science*, 265:2037 (1994)). Genome-wide LD studies involve using a dense collection of SNPs as markers to search for an ancestral haplotype carrying a disease-susceptibility allele. Such studies cannot be undertaken without the availability of an extremely dense SNP map and their potential for success
15 depends sensitively on many population genetic assumptions. Association studies are more straightforward because they directly test the hypothesis that a specific SNP increases disease risk. They make few assumptions, and require only the availability of a suitable database of appropriate SNPs. In the near term, focusing on cSNPs is likely to be most productive inasmuch as the class is easily recognized (in
20 contrast to regulatory polymorphisms) and is likely to contain a significant proportion of the disease-susceptibility alleles.

The present invention relates to a gene which comprises a single nucleotide polymorphism (SNP) at a specific location. The gene which includes the SNP has at least two alleles, referred to herein as the reference allele and the variant allele. The
25 reference allele (prototypical or wild type allele) has been designated arbitrarily and typically corresponds to the nucleotide sequence of the gene which has been deposited with GenBank or TIGR under a given Accession number. The variant allele differs from the reference allele by one at least one nucleotide at the site(s) identified in Figures 5A-5QQQQQQQ. The present invention also relates to variant
30 alleles of the described genes and to complements of the variant alleles. The invention further relates to portions of the variant alleles and portions of complements of the variant alleles which comprise (encompass) the site of the SNP

and are at least 5 nucleotides in length. Portions can be, for example, 5-10, 5-15, 10-20, 5-25, 10-30, 10-50 or 10-100 bases long. For example, a portion of a variant allele which is 5 nucleotides in length includes the single nucleotide polymorphism (the nucleotide which differs from the reference allele at that site) and four
5 additional nucleotides which flank the site in the variant allele. These nucleotides can be on one or both sides of the polymorphism. Polymorphisms which are the subject of this invention are defined in Figures 5A-5QQQQQQQ with respect to the reference sequence deposited in GenBank under the Accession number indicated. For example, the invention relates to a portion of a gene (e.g., AADC) having a
10 partial nucleotide sequence as shown in Figures 5A-5QQQQQQQ comprising a single nucleotide polymorphism at a specific position. The reference nucleotide for AADC is shown in column 3 and the variant nucleotide is shown in column 4 of Figures 5A-5QQQQQQQ. The nucleotide sequences of the invention can be double- or single-stranded.

15 The invention further provides allele-specific oligonucleotides that hybridize to a gene comprising a single nucleotide polymorphism or to the complement of the gene. These oligonucleotides can be probes or primers.

The invention further provides a method of analyzing a nucleic acid from an individual. The method determines which base is present at any one of the
20 polymorphic sites shown in Figures 5A-5QQQQQQQ. Optionally, a set of bases occupying a set of the polymorphic sites shown in Figures 5A-5QQQQQQQ is determined. This type of analysis can be performed on a number of individuals, who are tested for the presence of a disease phenotype. The presence or absence of disease phenotype is then correlated with a base or set of bases present at the
25 polymorphic site or sites in the individuals tested.

An oligonucleotide of this invention can be DNA or RNA, and single- or double-stranded. Oligonucleotides can be naturally occurring or synthetic, but are typically prepared by synthetic means. Preferred oligonucleotides of the invention include segments of DNA, or their complements, which include any one of the
30 polymorphic sites shown in Figures 5A-5QQQQQQQ. The segments can be between 5 and 250 bases, and, in specific embodiments, are between 5-10, 5-20, 10-20, 10-50, 20-50 or 10-100 bases. The polymorphic site can occur within any

position of the segment. The segments can be from any of the allelic forms of DNA shown in Figures 5A-5Q. Q. Q. Q. Q. Q. Q. Q. Q.

As used herein, the terms "nucleotide" and "nucleic acid" are intended to be equivalent. The terms "nucleotide sequence", "nucleic acid sequence", "nucleic acid molecule" and "segment" are intended to be equivalent.

Hybridization probes are oligonucleotides which bind in a base-specific manner to a complementary strand of nucleic acid. Such probes include peptide nucleic acids, as described in Nielsen *et al.*, *Science* 254, 1497-1500 (1991). Probes can be any length suitable for specific hybridization to the target nucleic acid sequence. The most appropriate length of the probe may vary depending upon the hybridization method in which it is being used; for example, particular lengths may be more appropriate for use in microfabricated arrays, while other lengths may be more suitable for use in classical hybridization methods. Suitable probes and primers can range from about 5 nucleotides to about 30 nucleotides in length. For example, probes and primers can be 5, 6, 8, 10, 12, 14, 16, 18, 20, 22, 24, 25, 26, 28 or 30 nucleotides in length. The probe or primer preferably contains at least one polymorphic site occupied by any of the possible variant nucleotides. The nucleotide sequence can correspond to the coding sequence of the allele or to the complement of the coding sequence of the allele.

As used herein, the term "primer" refers to a single-stranded oligonucleotide which acts as a point of initiation of template-directed DNA synthesis under appropriate conditions (*e.g.*, in the presence of four different nucleoside triphosphates and an agent for polymerization, such as, DNA or RNA polymerase or reverse transcriptase) in an appropriate buffer and at a suitable temperature. The appropriate length of a primer depends on the intended use of the primer, but typically ranges from 15 to 30 nucleotides. Short primer molecules generally require cooler temperatures to form sufficiently stable hybrid complexes with the template. A primer need not reflect the exact sequence of the template, but must be sufficiently complementary to hybridize with a template. The term primer site refers to the area of the target DNA to which a primer hybridizes. The term primer pair refers to a set of primers including a 5' (upstream) primer that hybridizes with the 5'

end of the DNA sequence to be amplified and a 3' (downstream) primer that hybridizes with the complement of the 3' end of the sequence to be amplified.

As used herein, linkage describes the tendency of genes, alleles, loci or genetic markers to be inherited together as a result of their location on the same chromosome. It can be measured by percent recombination between the two genes, alleles, loci or genetic markers.

As used herein, polymorphism refers to the occurrence of two or more genetically determined alternative sequences or alleles in a population. A polymorphic marker or site is the locus at which divergence occurs. Preferred markers have at least two alleles, each occurring at frequency of greater than 1%, and more preferably greater than 10% or 20% of a selected population. A polymorphic locus may be as small as one base pair. Polymorphic markers include restriction fragment length polymorphisms, variable number of tandem repeats (VNTR's), hypervariable regions, minisatellites, dinucleotide repeats, trinucleotide repeats, tetranucleotide repeats, simple sequence repeats, and insertion elements such as Alu. The first identified allelic form is arbitrarily designated as the reference form and other allelic forms are designated as alternative or variant alleles. The allelic form occurring most frequently in a selected population is sometimes referred to as the wildtype form. Diploid organisms may be homozygous or heterozygous for allelic forms. A diallelic or biallelic polymorphism has two forms. A triallelic polymorphism has three forms.

By altering amino acid sequence, SNPs may alter the function of the encoded proteins. The discovery of the SNP facilitates biochemical analysis of the variants and the development of assays to characterize the variants and to screen for pharmaceutical that would interact directly with on or another form of the protein. SNPs (including silent SNPs) may also alter the regulation of the gene at the transcriptional or post-transcriptional level. SNPs (including silent SNPs) also enable the development of specific DNA, RNA, or protein-based diagnostics that detect the presence or absence of the polymorphism in particular conditions.

A single nucleotide polymorphism occurs at a polymorphic site occupied by a single nucleotide, which is the site of variation between allelic sequences. The site

is usually preceded by and followed by highly conserved sequences of the allele (e.g., sequences that vary in less than 1/100 or 1/1000 members of the populations).

A single nucleotide polymorphism usually arises due to substitution of one nucleotide for another at the polymorphic site. A transition is the replacement of
5 one purine by another purine or one pyrimidine by another pyrimidine. A transversion is the replacement of a purine by a pyrimidine or vice versa. Single nucleotide polymorphisms can also arise from a deletion of a nucleotide or an insertion of a nucleotide relative to a reference allele. Typically the polymorphic site is occupied by a base other than the reference base. For example, where the
10 reference allele contains the base "T" at the polymorphic site, the altered allele can contain a "C", "G" or "A" at the polymorphic site.

Hybridizations are usually performed under stringent conditions, for example, at a salt concentration of no more than 1 M and a temperature of at least 25°C. For example, conditions of 5X SSPE (750 mM NaCl, 50 mM NaPhosphate,
15 5 mM EDTA, pH 7.4) and a temperature of 25-30°C, or equivalent conditions, are suitable for allele-specific probe hybridizations. Equivalent conditions can be determined by varying one or more of the parameters given as an example, as known in the art, while maintaining a similar degree of identity or similarity between the target nucleotide sequence and the primer or probe used.

20 The term "isolated" is used herein to indicate that the material in question exists in a physical milieu distinct from that in which it occurs in nature. For example, an isolated nucleic acid of the invention may be substantially isolated with respect to the complex cellular milieu in which it naturally occurs. In some instances, the isolated material will form part of a composition (for example, a crude
25 extract containing other substances), buffer system or reagent mix. In other circumstance, the material may be purified to essential homogeneity, for example as determined by PAGE or column chromatography such as HPLC. Preferably, an isolated nucleic acid comprises at least about 50, 80 or 90 percent (on a molar basis) of all macromolecular species present.

I. Analysis of Polymorphisms

A. Preparation of Samples

Polymorphisms are detected in a target nucleic acid from an individual being analyzed. For assay of genomic DNA, virtually any biological sample (other than
5 pure red blood cells) is suitable. For example, convenient tissue samples include whole blood, semen, saliva, tears, urine, fecal material, sweat, buccal, skin and hair. For assay of cDNA or mRNA, the tissue sample must be obtained from an organ in which the target nucleic acid is expressed. For example, if the target nucleic acid is a cytochrome P450, the liver is a suitable source.

10 Many of the methods described below require amplification of DNA from target samples. This can be accomplished by e.g., PCR. *See generally PCR Technology: Principles and Applications for DNA Amplification* (ed. H.A. Erlich, Freeman Press, NY, NY, 1992); *PCR Protocols: A Guide to Methods and Applications* (eds. Innis, *et al.*, Academic Press, San Diego, CA, 1990); Mattila *et al.*, *Nucleic Acids Res.* 19, 4967 (1991); Eckert *et al.*, *PCR Methods and Applications* 1, 17 (1991); *PCR* (eds. McPherson *et al.*, IRL Press, Oxford); and U.S. Patent 4,683,202.

Other suitable amplification methods include the ligase chain reaction (LCR) (see Wu and Wallace, *Genomics* 4, 560 (1989), Landegren *et al.*, *Science* 241, 1077
20 (1988), transcription amplification (Kwoh *et al.*, *Proc. Natl. Acad. Sci. USA* 86, 1173 (1989)), and self-sustained sequence replication (Guatelli *et al.*, *Proc. Nat. Acad. Sci. USA*, 87, 1874 (1990)) and nucleic acid based sequence amplification (NASBA). The latter two amplification methods involve isothermal reactions based on isothermal transcription, which produce both single stranded RNA (ssRNA) and
25 double stranded DNA (dsDNA) as the amplification products in a ratio of about 30 or 100 to 1, respectively.

B. Detection of Polymorphisms in Target DNA

There are two distinct types of analysis of target DNA for detecting polymorphisms. The first type of analysis, sometimes referred to as de novo
30 characterization, is carried out to identify polymorphic sites not previously characterized (i.e., to identify new polymorphisms). This analysis compares target

sequences in different individuals to identify points of variation, i.e., polymorphic sites. By analyzing groups of individuals representing the greatest ethnic diversity among humans and greatest breed and species variety in plants and animals, patterns characteristic of the most common alleles/haplotypes of the locus can be identified, and the frequencies of such alleles/haplotypes in the population can be determined. Additional allelic frequencies can be determined for subpopulations characterized by criteria such as geography, race, or gender. The de novo identification of polymorphisms of the invention is described in the Examples section. The second type of analysis determines which form(s) of a characterized (known) polymorphism are present in individuals under test. There are a variety of suitable procedures, which are discussed in turn.

1. Allele-Specific Probes

The design and use of allele-specific probes for analyzing polymorphisms is described by e.g., Saiki *et al.*, *Nature* 324, 163-166 (1986); Dattagupta, EP 235,726, Saiki, WO 89/11548. Allele-specific probes can be designed that hybridize to a segment of target DNA from one individual but do not hybridize to the corresponding segment from another individual due to the presence of different polymorphic forms in the respective segments from the two individuals. Hybridization conditions should be sufficiently stringent that there is a significant difference in hybridization intensity between alleles, and preferably an essentially binary response, whereby a probe hybridizes to only one of the alleles. Some probes are designed to hybridize to a segment of target DNA such that the polymorphic site aligns with a central position (e.g., in a 15-mer at the 7 position; in a 16-mer, at either the 8 or 9 position) of the probe. This design of probe achieves good discrimination in hybridization between different allelic forms.

Allele-specific probes are often used in pairs, one member of a pair showing a perfect match to a reference form of a target sequence and the other member showing a perfect match to a variant form. Several pairs of probes can then be immobilized on the same support for simultaneous analysis of multiple polymorphisms within the same target sequence.

2. Tiling Arrays

The polymorphisms can also be identified by hybridization to nucleic acid arrays, some examples of which are described in WO 95/11995. One form of such arrays is described in the Examples section in connection with de novo identification
5 of polymorphisms. The same array or a different array can be used for analysis of characterized polymorphisms. WO 95/11995 also describes subarrays that are optimized for detection of a variant form of a precharacterized polymorphism. Such a subarray contains probes designed to be complementary to a second reference sequence, which is an allelic variant of the first reference sequence. The second
10 group of probes is designed by the same principles as described in the Examples, except that the probes exhibit complementarity to the second reference sequence. The inclusion of a second group (or further groups) can be particularly useful for analyzing short subsequences of the primary reference sequence in which multiple mutations are expected to occur within a short distance commensurate with the
15 length of the probes (e.g., two or more mutations within 9 to 21 bases).

3. Allele-Specific Primers

An allele-specific primer hybridizes to a site on target DNA overlapping a polymorphism and only primes amplification of an allelic form to which the primer exhibits perfect complementarity. See Gibbs, *Nucleic Acid Res.* 17, 2427-2448
20 (1989). This primer is used in conjunction with a second primer which hybridizes at a distal site. Amplification proceeds from the two primers, resulting in a detectable product which indicates the particular allelic form is present. A control is usually performed with a second pair of primers, one of which shows a single base mismatch at the polymorphic site and the other of which exhibits perfect
25 complementarity to a distal site. The single-base mismatch prevents amplification and no detectable product is formed. The method works best when the mismatch is included in the 3'-most position of the oligonucleotide aligned with the polymorphism because this position is most destabilizing to elongation from the primer (see, e.g., WO 93/22456).

4. Direct-Sequencing

The direct analysis of the sequence of polymorphisms of the present invention can be accomplished using either the dideoxy chain termination method or the Maxam Gilbert method (see Sambrook *et al.*, *Molecular Cloning, A Laboratory*
5 *Manual* (2nd Ed., CSHP, New York 1989); Zyskind *et al.*, *Recombinant DNA Laboratory Manual*, (Acad. Press, 1988)).

5. Denaturing Gradient Gel Electrophoresis

Amplification products generated using the polymerase chain reaction can be analyzed by the use of denaturing gradient gel electrophoresis. Different alleles can
10 be identified based on the different sequence-dependent melting properties and electrophoretic migration of DNA in solution. Erlich, ed., *PCR Technology, Principles and Applications for DNA Amplification*, (W.H. Freeman and Co, New York, 1992), Chapter 7.

6. Single-Strand Conformation Polymorphism Analysis

15 Alleles of target sequences can be differentiated using single-strand conformation polymorphism analysis, which identifies base differences by alteration in electrophoretic migration of single stranded PCR products, as described in Orita *et al.*, *Proc. Nat. Acad. Sci.* 86, 2766-2770 (1989). Amplified PCR products can be generated as described above, and heated or otherwise denatured, to form single
20 stranded amplification products. Single-stranded nucleic acids may refold or form secondary structures which are partially dependent on the base sequence. The different electrophoretic mobilities of single-stranded amplification products can be related to base-sequence differences between alleles of target sequences.

II. Methods of Use

25 After determining polymorphic form(s) present in an individual at one or more polymorphic sites, this information can be used in a number of methods.

A. Forensics

Determination of which polymorphic forms occupy a set of polymorphic sites in an individual identifies a set of polymorphic forms that distinguishes the individual. See generally National Research Council, *The Evaluation of Forensic*
5 *DNA Evidence* (Eds. Pollard *et al.*, National Academy Press, DC, 1996). The more sites that are analyzed, the lower the probability that the set of polymorphic forms in one individual is the same as that in an unrelated individual. Preferably, if multiple sites are analyzed, the sites are unlinked. Thus, polymorphisms of the invention are often used in conjunction with polymorphisms in distal genes. Preferred
10 polymorphisms for use in forensics are biallelic because the population frequencies of two polymorphic forms can usually be determined with greater accuracy than those of multiple polymorphic forms at multi-allelic loci.

The capacity to identify a distinguishing or unique set of forensic markers in an individual is useful for forensic analysis. For example, one can determine
15 whether a blood sample from a suspect matches a blood or other tissue sample from a crime scene by determining whether the set of polymorphic forms occupying selected polymorphic sites is the same in the suspect and the sample. If the set of polymorphic markers does not match between a suspect and a sample, it can be concluded (barring experimental error) that the suspect was not the source of the
20 sample. If the set of markers does match, one can conclude that the DNA from the suspect is consistent with that found at the crime scene. If frequencies of the polymorphic forms at the loci tested have been determined (e.g., by analysis of a suitable population of individuals), one can perform a statistical analysis to determine the probability that a match of suspect and crime scene sample would
25 occur by chance.

$p(ID)$ is the probability that two random individuals have the same polymorphic or allelic form at a given polymorphic site. In biallelic loci, four genotypes are possible: AA, AB, BA, and BB. If alleles A and B occur in a haploid genome of the organism with frequencies x and y , the probability of each genotype
30 in a diploid organism is (see WO 95/12607):

Homozygote: $p(AA) = x^2$

Homozygote: $p(BB) = y^2 = (1-x)^2$

Single Heterozygote: $p(AB) = p(BA) = xy = x(1-x)$

Both Heterozygotes: $p(AB+BA) = 2xy = 2x(1-x)$

The probability of identity at one locus (i.e., the probability that two individuals, picked at random from a population will have identical polymorphic forms at a given locus) is given by the equation:

$$p(ID) = (x^2)^2 + (2xy)^2 + (y^2)^2.$$

These calculations can be extended for any number of polymorphic forms at a given locus. For example, the probability of identity $p(ID)$ for a 3-allele system where the alleles have the frequencies in the population of x , y and z , respectively, is equal to the sum of the squares of the genotype frequencies:

$$p(ID) = x^4 + (2xy)^2 + (2yz)^2 + (2xz)^2 + z^4 + y^4$$

In a locus of n alleles, the appropriate binomial expansion is used to calculate $p(ID)$ and $p(exc)$.

The cumulative probability of identity ($\text{cum } p(ID)$) for each of multiple unlinked loci is determined by multiplying the probabilities provided by each locus.

$$\text{cum } p(ID) = p(ID1)p(ID2)p(ID3).... p(IDn)$$

The cumulative probability of non-identity for n loci (i.e., the probability that two random individuals will be different at 1 or more loci) is given by the equation:

$$\text{cum } p(\text{nonID}) = 1 - \text{cum } p(ID).$$

If several polymorphic loci are tested, the cumulative probability of non-identity for random individuals becomes very high (e.g., one billion to one). Such probabilities can be taken into account together with other evidence in determining the guilt or innocence of the suspect.

B. Paternity Testing

The object of paternity testing is usually to determine whether a male is the father of a child. In most cases, the mother of the child is known and thus, the mother's contribution to the child's genotype can be traced. Paternity testing investigates whether the part of the child's genotype not attributable to the mother is consistent with that of the putative father. Paternity testing can be performed by analyzing sets of polymorphisms in the putative father and the child.

If the set of polymorphisms in the child attributable to the father does not match the set of polymorphisms of the putative father, it can be concluded, barring experimental error, that the putative father is not the real father. If the set of polymorphisms in the child attributable to the father does match the set of polymorphisms of the putative father, a statistical calculation can be performed to determine the probability of coincidental match.

The probability of parentage exclusion (representing the probability that a random male will have a polymorphic form at a given polymorphic site that makes him incompatible as the father) is given by the equation (see WO 95/12607):

$$p(\text{exc}) = xy(1-xy)$$

where x and y are the population frequencies of alleles A and B of a biallelic polymorphic site.

(At a triallelic site $p(\text{exc}) = xy(1-xy) + yz(1-yz) + xz(1-xz) + 3xyz(1-xyz)$), where x, y and z are the respective population frequencies of alleles A, B and C).

The probability of non-exclusion is

$$p(\text{non-exc}) = 1 - p(\text{exc})$$

The cumulative probability of non-exclusion (representing the value obtained when n loci are used) is thus:

$$\text{cum } p(\text{non-exc}) = p(\text{non-exc1})p(\text{non-exc2})p(\text{non-exc3})\dots p(\text{non-excn})$$

The cumulative probability of exclusion for n loci (representing the probability that a random male will be excluded)

$$\text{cum } p(\text{exc}) = 1 - \text{cum } p(\text{non-exc}).$$

If several polymorphic loci are included in the analysis, the cumulative probability of exclusion of a random male is very high. This probability can be taken into account in assessing the liability of a putative father whose polymorphic marker set matches the child's polymorphic marker set attributable to his/her father.

C. Correlation of Polymorphisms with Phenotypic Traits

The polymorphisms of the invention may contribute to the phenotype of an organism in different ways. Some polymorphisms occur within a protein coding sequence and contribute to phenotype by affecting protein structure. The effect may be neutral, beneficial or detrimental, or both beneficial and detrimental, depending

on the circumstances. For example, a heterozygous sickle cell mutation confers resistance to malaria, but a homozygous sickle cell mutation is usually lethal. Other polymorphisms occur in noncoding regions but may exert phenotypic effects indirectly via influence on replication, transcription, and translation. A single
5 polymorphism may affect more than one phenotypic trait. Likewise, a single phenotypic trait may be affected by polymorphisms in different genes. Further, some polymorphisms predispose an individual to a distinct mutation that is causally related to a certain phenotype.

Phenotypic traits include diseases that have known but hitherto unmapped
10 genetic components. Phenotypic traits also include symptoms of, or susceptibility to, multifactorial diseases of which a component is or may be genetic, such as autoimmune diseases, inflammation, cancer, diseases of the nervous system, and infection by pathogenic microorganisms. Some examples of diseases which can be treated or diagnosed as described herein include, but are not limited to,
15 bradyarrhythmias, tachyarrhythmias, heart failure, such as congestive heart failure, congenital heart disease, rheumatic fever, valvular heart disease, cardiomyopathies, myocarditides, pericardial diseases, cardiac tumors, cardiac manifestations of systemic diseases, and traumatic cardiac injury. Other disorders include atherosclerosis, acute myocardial infarction, ischemic heart disease, hypertensive
20 vascular disease, disorders of the aorta, vascular diseases of the extremities, vessel wall disorders, such as various forms of thrombocytopenia, von Willebrand's disease and drug-induced platelet dysfunction, and homeostatic disorders relating to vessel disease and associated bleeding. Also suitable are thrombotic thrombocytopenic purpura, hemolytic-uremic syndrome, Henoch-Schönlein purpura, capillary fragility,
25 vascular purpura, metabolic and inflammatory disorders, such as those induced by rickettsiae and certain drugs, such as sulfonamides, aortic aneurysm, aortic dissection, aortic occlusion, aortitis, atherosclerosis, coronary artery disease, angina, myocardial infarction, thrombosis, hemostatic and coagulation disorders, hypertension and hypotension. Other disorders include transplant accelerated
30 vascular restenosis following balloon angioplasty, Raynaud's disease and acrocyanosis.

Additional disorders include, but are not limited to, disorders of neurodegeneration characterized by astrocyte hypertrophy including gliosis, Pick's disease, aceroplasminemia, portal-systemic encephalopathy, frontal lobe dementia and inherited and acquired ataxias, neurodegenerative diseases of other etiology
5 including progressive supranuclear palsy, primary progressive aphasia, cortical basal degeneration, Alzheimer's disease, Huntington's disease, and Parkinson's disease, retinitis pigmentosa and amyotrophic lateral sclerosis. Other disorders include epilepsy, stroke, defects of neural migration and differentiation, including Miller-Dieker lissencephaly syndrome, and cancer of the brain including astrocytomas and
10 gliomas, as well as psychological disorders such as schizophrenia.

Phenotypic traits also include characteristics such as longevity, appearance (e.g., baldness, obesity), strength, speed, endurance, fertility, and susceptibility or receptivity to particular drugs or therapeutic treatments.

The correlation of one or more polymorphisms with phenotypic traits can be
15 facilitated by knowledge of the gene product of the wild type (reference) gene. The genes in which cSNPs of the present invention have been identified are genes which have been previously sequenced and characterized in one of their allelic forms.

Correlation is performed for a population of individuals who have been tested for the presence or absence of a phenotypic trait of interest and for
20 polymorphic markers sets. To perform such analysis, the presence or absence of a set of polymorphisms (i.e. a polymorphic set) is determined for a set of the individuals, some of whom exhibit a particular trait, and some of which exhibit lack of the trait. The alleles of each polymorphism of the set are then reviewed to determine whether the presence or absence of a particular allele is associated with
25 the trait of interest. Correlation can be performed by standard statistical methods such as a K-squared test and statistically significant correlations between polymorphic form(s) and phenotypic characteristics are noted. For example, it might be found that the presence of allele A1 at polymorphism A correlates with heart disease. As a further example, it might be found that the combined presence of
30 allele A1 at polymorphism A and allele B1 at polymorphism B correlates with increased milk production of a farm animal.

Such correlations can be exploited in several ways. In the case of a strong correlation between a set of one or more polymorphic forms and a disease for which treatment is available, detection of the polymorphic form set in a human or animal patient may justify immediate administration of treatment, or at least the institution of regular monitoring of the patient. Detection of a polymorphic form correlated with serious disease in a couple contemplating a family may also be valuable to the couple in their reproductive decisions. For example, the female partner might elect to undergo in vitro fertilization to avoid the possibility of transmitting such a polymorphism from her husband to her offspring. In the case of a weaker, but still statistically significant correlation between a polymorphic set and human disease, immediate therapeutic intervention or monitoring may not be justified. Nevertheless, the patient can be motivated to begin simple life-style changes (e.g., diet, exercise) that can be accomplished at little cost to the patient but confer potential benefits in reducing the risk of conditions to which the patient may have increased susceptibility by virtue of variant alleles. Identification of a polymorphic set in a patient correlated with enhanced receptiveness to one of several treatment regimes for a disease indicates that this treatment regime should be followed.

For animals and plants, correlations between characteristics and phenotype are useful for breeding for desired characteristics. For example, Beitz *et al.*, US 5,292,639 discuss use of bovine mitochondrial polymorphisms in a breeding program to improve milk production in cows. To evaluate the effect of mtDNA D-loop sequence polymorphism on milk production, each cow was assigned a value of 1 if variant or 0 if wildtype with respect to a prototypical mitochondrial DNA sequence at each of 17 locations considered. Each production trait was analyzed individually with the following animal model:

$$Y_{ijkpn} = \mu + YS_i + P_j + X_k + \beta_1 + \dots \beta_{17} + PE_n + a_n + e_p$$

where Y_{ijkpn} is the milk, fat, fat percentage, SNF, SNF percentage, energy concentration, or lactation energy record; μ is an overall mean; YS_i is the effect common to all cows calving in year-season; X_k is the effect common to cows in either the high or average selection line; β_1 to β_{17} are the binomial regressions of production record on mtDNA D-loop sequence polymorphisms; PE_n is permanent environmental effect common to all records of cow n ; a_n is effect of animal n and is

composed of the additive genetic contribution of sire and dam breeding values and a Mendelian sampling effect; and e_p is a random residual. It was found that eleven of seventeen polymorphisms tested influenced at least one production trait. Bovines having the best polymorphic forms for milk production at these eleven loci are used
5 as parents for breeding the next generation of the herd.

D. Genetic Mapping of Phenotypic Traits

The previous section concerns identifying correlations between phenotypic traits and polymorphisms that directly or indirectly contribute to those traits. The present section describes identification of a physical linkage between a genetic locus
10 associated with a trait of interest and polymorphic markers that are not associated with the trait, but are in physical proximity with the genetic locus responsible for the trait and co-segregate with it. Such analysis is useful for mapping a genetic locus associated with a phenotypic trait to a chromosomal position, and thereby cloning gene(s) responsible for the trait. See Lander *et al.*, *Proc. Natl. Acad. Sci. (USA)* 83,
15 7353-7357 (1986); Lander *et al.*, *Proc. Natl. Acad. Sci. (USA)* 84, 2363-2367 (1987); Donis-Keller *et al.*, *Cell* 51, 319-337 (1987); Lander *et al.*, *Genetics* 121, 185-199 (1989)). Genes localized by linkage can be cloned by a process known as directional cloning. See Wainwright, *Med. J. Australia* 159, 170-174 (1993); Collins, *Nature Genetics* 1, 3-6 (1992).

20 Linkage studies are typically performed on members of a family. Available members of the family are characterized for the presence or absence of a phenotypic trait and for a set of polymorphic markers. The distribution of polymorphic markers in an informative meiosis is then analyzed to determine which polymorphic markers co-segregate with a phenotypic trait. See, *e.g.*, Kerem *et al.*, *Science* 245, 1073-1080
25 (1989); Monaco *et al.*, *Nature* 316, 842 (1985); Yamoka *et al.*, *Neurology* 40, 222-226 (1990); Rossiter *et al.*, *FASEB Journal* 5, 21-27 (1991).

Linkage is analyzed by calculation of LOD (log of the odds) values. A lod value is the relative likelihood of obtaining observed segregation data for a marker and a genetic locus when the two are located at a recombination fraction θ , versus
30 the situation in which the two are not linked, and thus segregating independently (Thompson & Thompson, *Genetics in Medicine* (5th ed, W.B. Saunders Company,

Philadelphia, 1991); Strachan, "Mapping the human genome" in *The Human Genome* (BIOS Scientific Publishers Ltd, Oxford), Chapter 4). A series of likelihood ratios are calculated at various recombination fractions (θ), ranging from $\theta = 0.0$ (coincident loci) to $\theta = 0.50$ (unlinked). Thus, the likelihood at a given value of θ is: probability of data if loci linked at θ to probability of data if loci
5 unlinked. The computed likelihoods are usually expressed as the \log_{10} of this ratio (i.e., a lod score). For example, a lod score of 3 indicates 1000:1 odds against an apparent observed linkage being a coincidence. The use of logarithms allows data collected from different families to be combined by simple addition. Computer
10 programs are available for the calculation of lod scores for differing values of θ (e.g., LIPED, MLINK (Lathrop, *Proc. Nat. Acad. Sci. (USA)* 81, 3443-3446 (1984)). For any particular lod score, a recombination fraction may be determined from mathematical tables. See Smith *et al.*, *Mathematical tables for research workers in human genetics* (Churchill, London, 1961); Smith, *Ann. Hum. Genet.* 32, 127-150
15 (1968). The value of θ at which the lod score is the highest is considered to be the best estimate of the recombination fraction.

Positive lod score values suggest that the two loci are linked, whereas negative values suggest that linkage is less likely (at that value of θ) than the possibility that the two loci are unlinked. By convention, a combined lod score of
20 +3 or greater (equivalent to greater than 1000:1 odds in favor of linkage) is considered definitive evidence that two loci are linked. Similarly, by convention, a negative lod score of -2 or less is taken as definitive evidence against linkage of the two loci being compared. Negative linkage data are useful in excluding a chromosome or a segment thereof from consideration. The search focuses on the
25 remaining non-excluded chromosomal locations.

III. Modified Polypeptides and Gene Sequences

The invention further provides variant forms of nucleic acids and corresponding proteins. The nucleic acids comprise one of the sequences described in Figures 5A-5Q, column 11, in which the polymorphic position is
30 occupied by one of the alternative bases for that position. Some nucleic acids encode full-length variant forms of proteins. Similarly, variant proteins have the

prototypical amino acid sequences encoded by nucleic acid sequences shown in Figures 5A-5QQQQQQQ, column 11, (read so as to be in-frame with the full-length coding sequence of which it is a component) except at an amino acid encoded by a codon including one of the polymorphic positions shown in Figures 5A-

5 5QQQQQQQ. That position is occupied by the amino acid coded by the corresponding codon in any of the alternative forms shown in Figures 5A-5QQQQQQQ.

Variant genes can be expressed in an expression vector in which a variant gene is operably linked to a native or other promoter. Usually, the promoter is a
10 eukaryotic promoter for expression in a mammalian cell. The transcription regulation sequences typically include a heterologous promoter and optionally an enhancer which is recognized by the host. The selection of an appropriate promoter, for example trp, lac, phage promoters, glycolytic enzyme promoters and tRNA promoters, depends on the host selected. Commercially available expression vectors
15 can be used. Vectors can include host-recognized replication systems, amplifiable genes, selectable markers, host sequences useful for insertion into the host genome, and the like.

The means of introducing the expression construct into a host cell varies depending upon the particular construction and the target host. Suitable means
20 include fusion, conjugation, transfection, transduction, electroporation or injection, as described in Sambrook, *supra*. A wide variety of host cells can be employed for expression of the variant gene, both prokaryotic and eukaryotic. Suitable host cells include bacteria such as *E. coli*, yeast, filamentous fungi, insect cells, mammalian cells, typically immortalized, *e.g.*, mouse, CHO, human and monkey cell lines and
25 derivatives thereof. Preferred host cells are able to process the variant gene product to produce an appropriate mature polypeptide. Processing includes glycosylation, ubiquitination, disulfide bond formation, general post-translational modification, and the like.

The protein may be isolated by conventional means of protein biochemistry
30 and purification to obtain a substantially pure product, *i.e.*, 80, 95 or 99% free of cell component contaminants, as described in Jacoby, *Methods in Enzymology* Volume 104, Academic Press, New York (1984); Scopes, *Protein Purification, Principles*

and Practice, 2nd Edition, Springer-Verlag, New York (1987); and Deutscher (ed), *Guide to Protein Purification, Methods in Enzymology*, Vol. 182 (1990). If the protein is secreted, it can be isolated from the supernatant in which the host cell is grown. If not secreted, the protein can be isolated from a lysate of the host cells.

- 5 The invention further provides transgenic nonhuman animals capable of expressing an exogenous variant gene and/or having one or both alleles of an endogenous variant gene inactivated. Expression of an exogenous variant gene is usually achieved by operably linking the gene to a promoter and optionally an enhancer, and microinjecting the construct into a zygote. *See Hogan et al.*,
10 "Manipulating the Mouse Embryo, A Laboratory Manual," Cold Spring Harbor Laboratory. Inactivation of endogenous variant genes can be achieved by forming a transgene in which a cloned variant gene is inactivated by insertion of a positive selection marker. *See Capecchi, Science* 244, 1288-1292 (1989). The transgene is then introduced into an embryonic stem cell, where it undergoes homologous
15 recombination with an endogenous variant gene. Mice and other rodents are preferred animals. Such animals provide useful drug screening systems.

 In addition to substantially full-length polypeptides expressed by variant genes, the present invention includes biologically active fragments of the polypeptides, or analogs thereof, including organic molecules which simulate the
20 interactions of the peptides. Biologically active fragments include any portion of the full-length polypeptide which confers a biological function on the variant gene product, including ligand binding, and antibody binding. Ligand binding includes binding by nucleic acids, proteins or polypeptides, small biologically active molecules, or large cellular structures.

- 25 Polyclonal and/or monoclonal antibodies that specifically bind to variant gene products but not to corresponding prototypical gene products are also provided. Antibodies can be made by injecting mice or other animals with the variant gene product or synthetic peptide fragments thereof. Monoclonal antibodies are screened as are described, for example, in Harlow & Lane, *Antibodies, A Laboratory Manual*,
30 Cold Spring Harbor Press, New York (1988); Goding, *Monoclonal antibodies, Principles and Practice* (2d ed.) Academic Press, New York (1986). Monoclonal antibodies are tested for specific immunoreactivity with a variant gene product and

lack of immunoreactivity to the corresponding prototypical gene product. These antibodies are useful in diagnostic assays for detection of the variant form, or as an active ingredient in a pharmaceutical composition.

IV. Kits

5 The invention further provides kits comprising at least one allele-specific oligonucleotide as described above. Often, the kits contain one or more pairs of allele-specific oligonucleotides hybridizing to different forms of a polymorphism. In some kits, the allele-specific oligonucleotides are provided immobilized to a substrate. For example, the same substrate can comprise allele-specific
10 oligonucleotide probes for detecting at least 10, 100 or all of the polymorphisms shown in Figures 5A-5Q. Optional additional components of the kit include, for example, restriction enzymes, reverse-transcriptase or polymerase, the substrate nucleoside triphosphates, means used to label (for example, an avidin-enzyme conjugate and enzyme substrate and chromogen if the label is biotin), and
15 the appropriate buffers for reverse transcription, PCR, or hybridization reactions. Usually, the kit also contains instructions for carrying out the methods.

From the foregoing, it is apparent that the invention includes a number of general uses that can be expressed concisely as follows. The invention provides for the use of any of the nucleic acid segments described above in the diagnosis or
20 monitoring of diseases, such as coronary artery disease, diabetes, coagulation disorders, lipid metabolism disorders, energy metabolism disorders, diseases of the blood, blood vessels and cardiovascular system, and infection by microorganisms, as well as psychological disorders (e.g., bipolar disorder, schizophrenia). The invention further provides for the use of any of the nucleic acid segments in the
25 manufacture of a medicament for the treatment or prophylaxis of such diseases. The invention further provides for the use of any of the DNA segments as a pharmaceutical.

While this invention has been particularly shown and described with references to preferred embodiments thereof, it will be understood by those skilled
30 in the art that various changes in form and details may be made therein without

departing from the spirit and scope of the invention as defined by the appended claims.

CLAIMS

What is claimed is:

1. A nucleic acid molecule selected from the group consisting of the genes listed in Figures 5A-5QQQQQQQ, wherein said nucleic acid molecule is at least 5 nucleotides in length and comprises a polymorphic site identified in Figures 5A-5QQQQQQQ, wherein a nucleotide at the polymorphic site is different from a nucleotide at the polymorphic site in a corresponding reference allele.
2. A nucleic acid molecule according to Claim 1, wherein said nucleic acid molecule is at least 10 nucleotides in length.
3. A nucleic acid molecule according to Claim 1, wherein said nucleic acid molecule is at least 20 nucleotides in length.
4. A nucleic acid molecule according to Claim 1, wherein the nucleotide at the polymorphic site is the variant nucleotide for the gene listed in Figures 5A-5QQQQQQQ.
5. An allele-specific oligonucleotide that hybridizes to a portion of a gene selected from the group consisting of the genes listed in Figures 5A-5QQQQQQQ, wherein said portion is at least 5 nucleotides in length and comprises a polymorphic site identified in Figures 5A-5QQQQQQQ, wherein a nucleotide at the polymorphic site is different from a nucleotide at the polymorphic site in a corresponding reference allele.
6. An allele-specific oligonucleotide according to Claim 5 that is a probe.
7. An allele-specific oligonucleotide according to Claim 5, wherein a central position of the probe aligns with the polymorphic site of the portion.

8. An allele-specific oligonucleotide according to Claim 5 that is a primer.
9. An allele-specific oligonucleotide according to Claim 8, wherein the 3' end of the primer aligns with the polymorphic site of the portion.
10. An isolated gene product encoded by a nucleic acid molecule according to
5 Claim 1.
11. A method of analyzing a nucleic acid sample, comprising obtaining the nucleic acid from an individual sample; and determining a base occupying any one of the polymorphic sites shown in Figures 5A-5QQQQQQQ.
12. A method according to Claim 11, wherein the nucleic acid sample is obtained
10 from a plurality of individuals, and a base occupying one of the polymorphic positions is determined in each of the individuals, and the method further comprising testing each individual for the presence of a disease phenotype, and correlating the presence of the disease phenotype with the base.

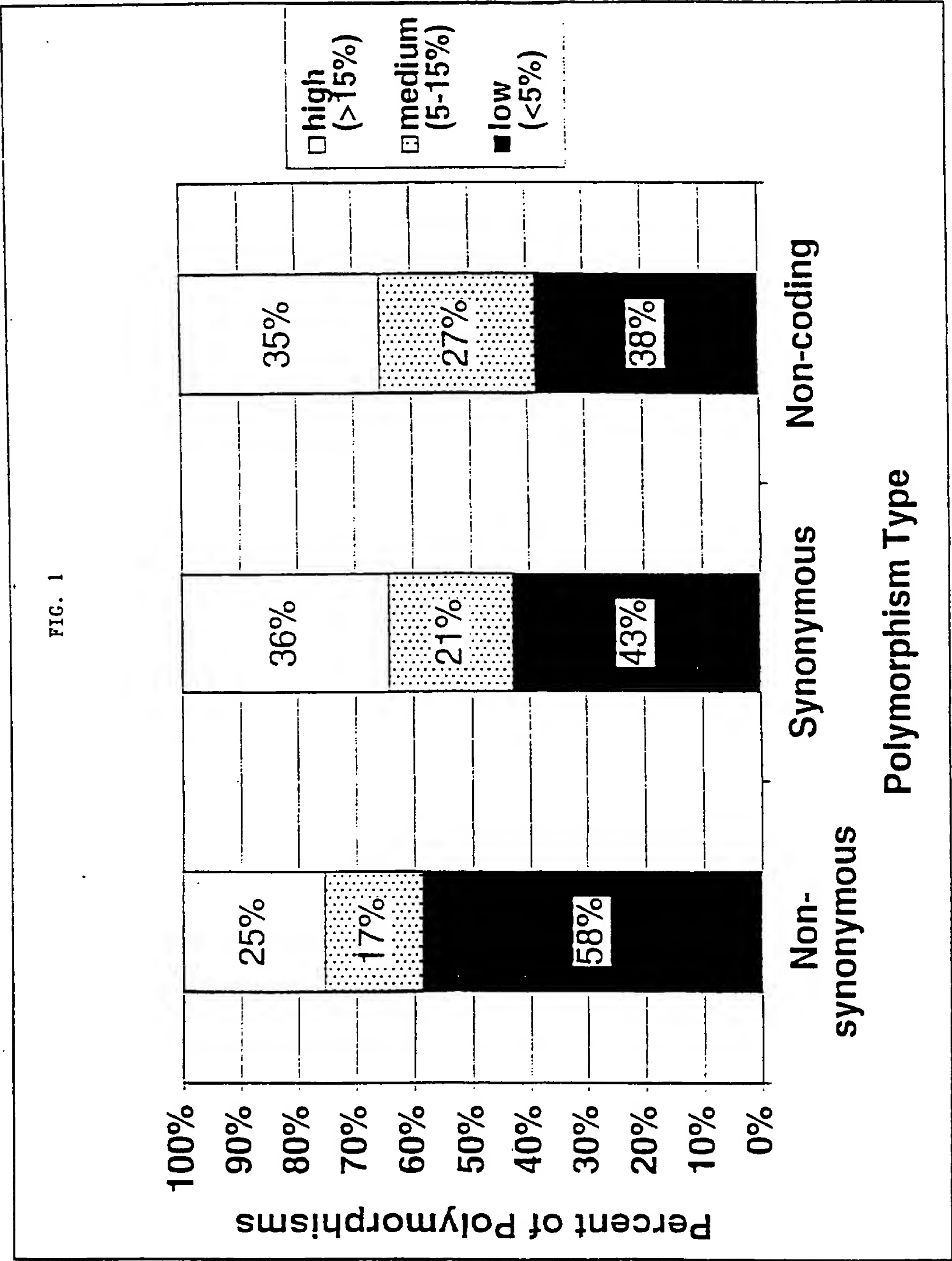
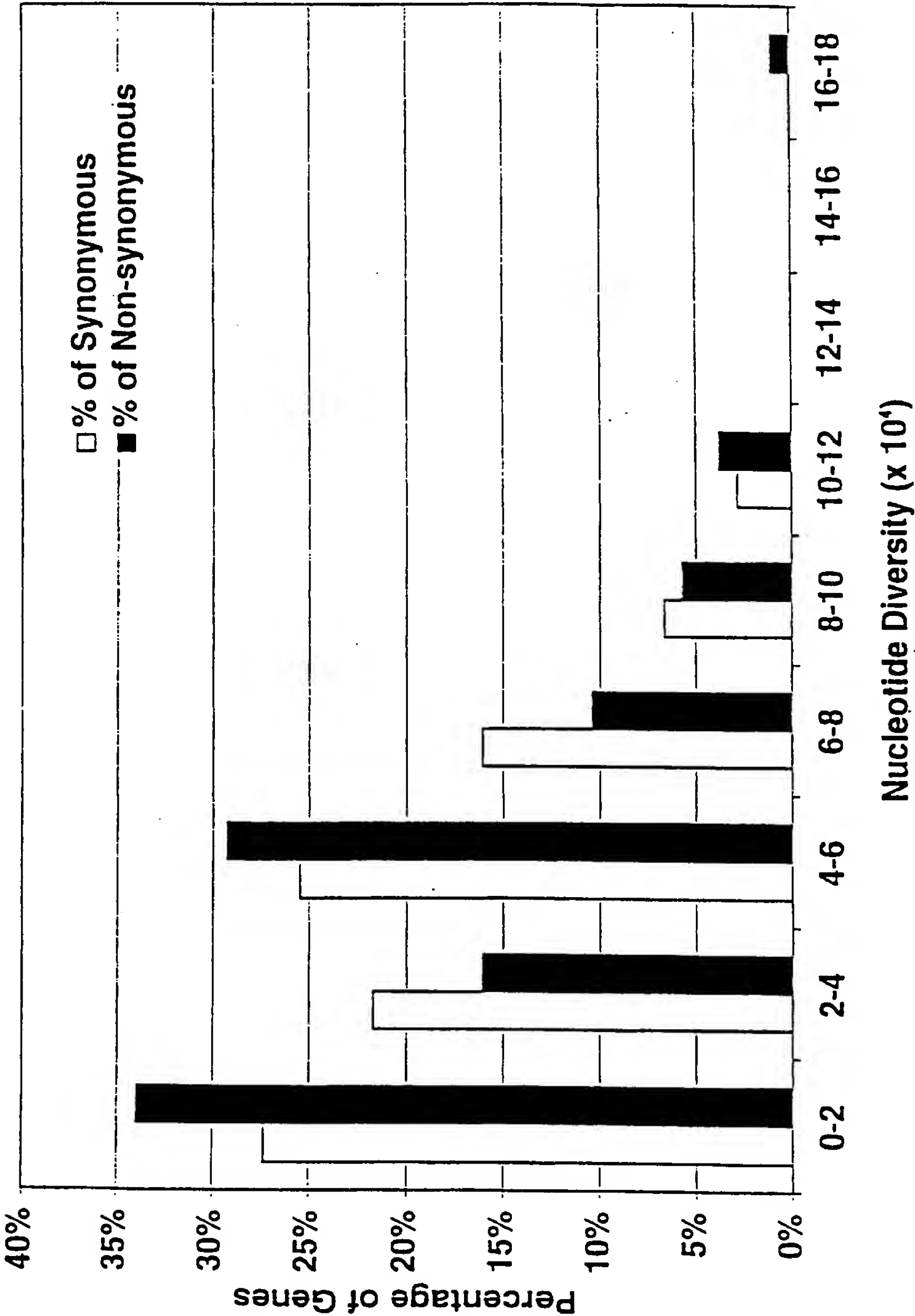


FIG. 2



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Gene	coding bp screened	No. Synonymous polymorphisms	No. Non- synonymous polymorphisms	Non-coding bp screened	No. Non-coding polymorphisms
AADC	1229	0	2	311	0
ADORA2	332	0	1	75	0
AHC	1413	0	0	63	1
ANX3	929	2	4	725	6
APOD	570	1	3	383	1
AR	2759	3	1	300	0
AT3	1357	3	0	121	0
BDNF	744	0	1	212	0
CD36	1209	1	1	252	0
CETP	1397	4	4	299	0
CGA	349	1	0	235	0
CLanalog	1461	3	2	12	0
CNTF	603	0	1	154	0
COMT	783	2	1	241	1
CRH	51	0	0	745	3
CYP11A	1556	1	1	547	0
CYP11B1	1410	7	7	496	9
CYP11B2	1512	7	8	906	4
CYP17	1395	3	0	36	0
CYP21	1488	5	11	542	7
DBH	1266	0	2	49	0
DRD1	1341	1	0	81	0
DRD2	1032	2	0	1379	3
DRD3	719	0	1	145	0
DRD5	1408	2	1	34	0
F10	1369	3	2	416	1
F11	1878	7	4	1312	2
F13A1	2199	3	6	948	4
F13B	1952	4	6	2339	4
F2	1740	3	2	292	0
F2R	1202	2	1	13	0
F3	875	0	1	92	0
F5	6564	13	16	1542	8
F7	1262	4	2	1209	2
F9	1364	0	1	1062	2
FGA	1935	2	2	490	0
FGB	1476	7	3	1057	0
FGG	1252	0	2	1392	2
FSH	355	1	1	44	0
FSHR	1683	1	3	0	0
GABRB1	1425	5	0	804	2
GAP43	675	1	1	79	0
GH1	644	0	1	426	5
GHR	1765	1	6	391	1
GNRHR	237	0	1	513	0
GP1BA	1881	2	2	48	0
GP1BB	1238	0	0	73	0
GP5	1683	0	0	52	0
GP9	534	1	0	143	0
GRF	224	0	0	239	0
GRIN1	1681	1	0	553	0
GRL	2334	4	3	4028	5
HCF2	1500	3	3	64	1

FIG. 3A

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Gene	coding bp screened	No. Synonymous polymorphisms	No. Non- synonymous polymorphisms	Non-coding bp screened	No. Non-coding polymorphisms
HMGCR	1724	0	1	12	1
HSD3B1	1122	3	2	653	1
HSD3B2	1122	1	1	723	2
HTR1A	1272	1	0	1189	1
HTR1D	1134	1	1	46	0
HTR1DB	1173	2	0	85	1
HTR1E	1098	1	1	70	0
HTR1EL	1101	1	0	46	0
HTR2A	1398	2	3	1709	9
HTR2C	1245	0	1	138	0
HTR5A	1062	2	0	34	0
HTR6	437	1	0	34	0
HTR7	1279	0	0	138	0
IGF1	630	0	0	7250	8
IGF2	546	0	0	610	1
ITGA2B	2833	4	3	707	0
ITGB3	2131	4	3	163	0
KLK2	297	0	1	279	2
LCAT	1289	1	2	90	0
LDLR	2101	7	3	38	0
LIPC	1471	4	3	754	4
LPL	409	1	1	48	0
MAOA	1032	1	0	69	0
MAOB	980	1	0	135	0
MPL	1748	1	2	903	1
NGFB	726	1	1	1186	5
NOS1	127	0	0	56	0
NT3	774	1	0	150	0
NTRK1	1961	5	2	1106	0
PACE	1500	2	0	1095	4
PAI1	1171	1	2	911	1
PAI2	1248	5	4	915	5
PC1	1881	1	3	456	1
PCI	1221	5	5	576	4
POMC	132	0	0	520	0
PRL	633	1	1	180	1
PROC	1334	3	0	114	0
PROS1	1868	1	0	557	0
PTAFR	1029	0	2	13	0
PTH	348	1	0	230	2
PTHLH	634	0	0	2342	13
SELP	2096	5	8	14	0
SHBG	1209	1	3	494	1
SLC6A1	1388	2	0	547	2
SLC6A3	1496	6	1	205	0
SLC6A4	1623	1	2	824	1
TBXA2R	1006	1	0	12	0
TBXAS1	1605	1	6	1411	1
TFPI	806	0	1	139	0
TH	965	1	1	104	0
THBD	1728	0	0	26	0
THPO	1049	0	0	632	2
VLDLR	2391	3	1	850	2
ALL GENES	135823	203	192	59552	150

FIG. 3B

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Polymorphism rates for different classes of sites. Nucleotide diversity and heterozygosity (π) are expressed x 10⁴.

Adjusted for frequency of sites*					
Polymorphism Type	bp screened	No. polys	Frequency (SNP/bp)	$\hat{\theta}$	π
Non-coding	59,552	150	1/397	4.93 ± 1.24	5.05 ± 2.40
Coding	135,823	395	1/344	5.47 ± 1.32	5.07 ± 2.40
synonymous		203	1/669	2.81 ± 0.68	2.98 ± 1.42
non-synonymous		192	1/707	2.66 ± 0.64	2.06 ± 0.98
conservative		122	1/1113	1.69 ± 0.41	1.44 ± 0.68
non-conservative		70	1/1940	0.97 ± 0.23	0.63 ± 0.30
four-fold degenerate sites	21,645	111	1/195	9.64 ± 2.32	9.26 ± 4.40
two-fold degenerate sites	34,294	125	1/274	6.85 ± 1.65	5.33 ± 2.53
non-degenerate sites	79,659	157	1/507	3.70 ± 0.89	2.52 ± 1.19
Total	195,375	545	1/357	5.31 ± 1.28	5.01 ± 2.38

* The number of synonymous sites was calculated as the sum of four-fold degenerate sites and half the number of two-fold degenerate sites; the number of non-synonymous sites is the sum of the non-degenerate sites and half the two-fold degenerate sites. The number of conservative and non-conservative sites is estimated as the proportion of non-synonymous sites at which a nucleotide substitution would create a conservative or non-conservative substitution, calculated as in footnote 21.

FIG. 4

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')
AADCd4	AADC	C	T	P	L	cds	GE1048	CACACACTGTACAATCCAA	CCTTACAAGAAAGGAATCAGGC
AADCd5	AADC	A	G	M	V	cds	GE1048	CACACACTGTACAATCCAA	CCTTACAAGAAAGGAATCAGGC
AADCd6	AADC	G	A	V	M	cds	GE1094	CACTGAAATCATTTTCTTTTCG	ACACACTTACC CCAGGC
AADCd7	AADC	C	T	D	D	cds	GE1263	CCCTTGTTACTGCTGACCCC	CACCTCTCCCC CTTCTC
AADCu1	AADC	C	T	R	W	cds	GE1094	CACTGAAATCATTTTCTTTTCG	ACACACTTACC CCAGGC
AADCu2	AADC	A	G	E	G	cds	GE1094	CACTGAAATCATTTTCTTTTCG	ACACACTTACC CCAGGC
AADCu3	AADC	A	T	I	I	cds	GE1004	TCCATCTGGGG ACTCAC	GTGCACCTPACC TCCACTC
ADORA ₂ u1	ADORA ₂	C	T	A	V	cds	GE1141	ATGGACCGTGA GCTGGC	AAGCCTGGSCA CCAACA
ADORA ₂ u2	ADORA ₂	C	G	P	P	cds	GE1141	ATGGACCGTGA GCTGGC	AAGCCTGGSCA CCAACA

FIG. 5A

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
AHCu5	AHC	A	G	Q	R	cds	GE654	TAAAGAGGCGC TACCAGGCG	TGAGCTGGGAA AAGCCG	TAAAGAGGCGCTACCAGGCGGgggggccacggcgctctctgtaccgcgtcgtctcttgcgggtgaag accacccgc [a/g] gcaggcagcacccctctactgcgtgccccacgagcacaaatcaagcgagggc ggctccggaggagcgccgagggcccccctgggtggacacacctctctggtgcgtgcgcgcggcgggtg cgctcaagaggtccacaggtggtctgcgagcgacccctcagcgggacctgtgaaagacgtcgcgtctc gtcaagtacttgccctgcttccagggtgctgccccctggaccagcagctgggtgctggtgcgcaactg ctggcgctccctgctcatgcttgagctggccacgagccgcttcgagttcgagactgtggaagtct cggagccacgcatgctcagaagatcctcacaccagcgccgggagaccggggggaacagagccca ctgcccgtgccccacgtgcagcaccatttggcacccggcgagggagggagaggtgccccccgcg ctcccaggtccaagccatcaagtgtcttcttccaaatgctggagctcgaacatcagttaccacagg agtacgctactcaaggggaccgtgctcttaccgccgggtaagggtacttgcccttagggcgcCGG CTTTTCCCAGCTCA	664
AHCu6	AHC	T	G	L	R	cds	GE654	TAAAGAGGCGC TACCAGGCG	TGAGCTGGGAA AAGCCG	TAAAGAGGCGCTACCAGGCGGgggggccacggcgctctctgtaccgcgtcgtcttgcgggtgaag accacccgcagcaggcagcacccctctactgcgtgccccacgagcacaaatcaagcgagggcgt ccggaggagcgccgagggcccccctgggtggacacacctctctggtgcgtgcgcgcggcgggtg caagagtcacaggtggtctgcgagcgacccctcagcgggacctgtgtgaagacgtcgcgtctcgta agtaactgcccgtctccagggtctgccccctggaccagcagctgggtgctgctgcgcaactgctgg ggctccctgctcatg [t/g] tgaagtcggccagggacccgttcgagttcgagactgtggaagtct cggagccacgcatgctcagaagatcctcacaccagcgccgggagaccggggggaacagagccca ctgcccgtgccccacgtgcagcaccatttggcacccggcgagggagggagaggtgccccccgcg ctcccaggtccaagccatcaagtgtcttcttccaaatgctggagctcgaacatcagttaccacagg agtacgctactcaaggggaccgtgctcttaccgccgggtaagggtacttgcccttagggcgcCGG CTTTTCCCAGCTCA	664
ANX3d1 5	ANX3	T	C	F	S	cds	GE444	TCTCTATGTTG CTTTGTGACCA AT	GACTTACTTTA ATTGCTGAATA TAGG	TCCTATGTTGTTGTGACCAATgacatttggltgtggaacacctgcattcccttaacagggtat tggaaactgatgagtttactctggaaccgaataatgggtgtccagatcagaataatgaccttttgaca ttcgaaacaggt [t/c] caagaagcatattggctctatccCTATATTCAGCAATTAAGTAAGTC	194
ANX3d1 6	ANX3	G	A	-	-	noncoding	GE444	TCTCTATGTTG CTTTGTGACCA AT	GACTTACTTTA ATTGCTGAATA TAGG	TCCTATGTTGTTGTGACCAATgacatttgt [g/a] ttgtgaaacacctgcattcccttaacagg gtattggaaactgatgagtttactctgaaaccgaataatgggtgtccagatcagaataatgaccttttg gacattcgaaacagaggttcaagaagcatltaggtctatccCTATATTCAGCAATTAAGTAAGTC	194
ANX3d1 7	ANX3	A	T	-	-	noncoding	GE453	TGTTGGCATTT AAACTTTTCTC	AAAAATTGTTA TTGAGCATGCA G	TGTTGGCATTTAAACTTTTCTCTggtttataataatatttttcagtttgtcaaggcttaatttca ttctgatttgggttcaagtatcagaagaagctcttggagatgacattagttccgaaacatctgggtg acttccggaaagctctgtgactttggcagatgtaaggttttattttttt [a/t] taactc cccagtaagCTGCATGCTCAATAACAATTTT	227
ANX3d1 8	ANX3	T	C	-	-	noncoding	GE433	TTACTTACTAT AGATTAAACCA ATTTC	TTTCCCAAGGG AATTAAAGG	TTACTTACTATAGATTAAACCAATTTCTattccgtgaagctgaacat [t/c] atttgccttttgt tacagttaatgtgtgaggaacacccggcctttttagccgaagaagctgcactgagccttgaagg ttggtctggaaggttcaatgtgcatctttagcgtCCCTTAATTCCTTGGGAAA	183
ANX3d1 9	ANX3	T	C	-	-	noncoding	GE439	TGATTTCATT TGGTCTCCCAT T	GAAGTAAGGTG GAGCTGTGG	TGATTTCATTATGTTCTCCCAATtatttatactgtatttggtttcaattgtatttcttttgcag tcggataactcttgagactatgaaatcacactcttaaaaactgtgtgtggagatgactgaacca gaagataactctccaaagggtccacgattgggt [t/c] CCAACAGCTCCACCTTACTTTC	189
ANX3d2 0	ANX3	T	C	-	-	noncoding	GE439	TGATTTCATT TGGTCTCCCAT T	GAAGTAAGGTG GAGCTGTGG	TGATTTCATTATGTTCTCCCAATtatttatactgtatttggtttcaattgtatttcttttgcag gcagtcggataactcttgagactatgaaatcacactcttaaaaactgtgtgtggagatgactga ccaagaagataactctccaaagggtccacgattgggttttCCAACAGCTCCACCTTACTTTC	189
ANX3u1	ANX3	T	C	-	-	noncoding	GE443	GATGTCATTT GNAACCAATG	TGTAACCTTCC TGATTTCCTTC	GATGTCATTTTGAACCAATGgacttttcaagtatttcttctagggtggacaccgaggaacagtaa gagattatccagactttagcccatcagtgagtgctgaagctattcgaagaagcaatccagggaatt ggtagtga [t/c] attttacaattcttcttcttaattgttGAAGCAAAATCAGGCAAGTTACA	191

FIG. 5C

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')
ANX3u9	ANX3	G	A	S	N	cds	GE443	GATGTCATTTTGAACCAATG	TGTAACTTGCC TGATTGGCTC
APODd7	APOD	T	C	F	S	cds	GEJ20	AGAAGGCTCAG AATGACAACC	CAGCATATAATTA CATTGAACCTT GTAG
APODd8	APOD	C	G	-	-	noncoding	GE371	CAGCCTTCTTG TGTGTCTCTG	TGCTGGTTGAT TGCTTTGTCT
APODd9	APOD	C	A	-	-	noncoding	GE371	CAGCCTTCTTG TGTGTCTCTG	TGCTGGTTGAT TGCTTTGTCT
APODu1	APOD	A	T	E	V	cds	GE304	CTGGCTCTGCA CGACAATG	ACCCAGTCACT CTGCCGTCA
APODu2	APOD	C	T	D	D	cds	GEJ20	AGAAGGCTCAG AATGACAACC	CAGCATATAATTA CATTGAACCTT GTAG
APODu3	APOD	T	G	S	A	cds	GE286	GCTTATTGGCT TTCAATGAGTT GT	TGCTCTCAGGAA TTCTCCAAGC
APODu4	APOD	C	T	S	L	cds	GE371	CAGCCTTCTTG TGTGTCTCTG	TGCTGGTTGAT TGCTTTGTCT

FIG. 5E

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
ANX3u10	ANX3	T	G	L	R	cds	GE447	AAAAGTATTTT CACATTTTTC C	TGATGACTTGG TCAAACCC	AAAAGTATTTTTCACATTTTTCCTCcttggttttggatttttagagc(t/g)gaagatgacttgaagggtgatctctctggccactttgagcatctcatggtgcccctagtgactcc(a/g)ceagcagctcttgatgcaagagcgtaaagaaatccatgaaggtatgagcccccaagccatttctgcccagggtttgaccaaagtcATCA
ANX3u11	ANX3	A	G	P	P	cds	GE447	AAAAGTATTTT CACATTTTTC C	TGATGACTTGG TCAAACCC	AAAAGTATTTTTCACATTTTTCCTCcttggttttggatttttagagcgtgaagatgacttgaagggttgatctctctggccactttgagcatctcatggtgcccctagtgactcc(a/g)ceagcagctcttgatgcaagagcgtaaagaaatccatgaaggtatgagcccccaagccatttctgcccagggtttgaccaaagtcATCA
ANX3u12	ANX3	T	G	L	R	cds	GE441	AAAAGAAATA ATTGTGCTCT AATATC	TGTAAACAA CATCAACACA AAGG	AAAAGAAATAATTGTGCTCTCTAATATATCattctctgtgatatagatctctctataaagctggtgagaaacagatggggcaggatgaagcaaatctcactgagatcc(t/g)gtgtttaaggagctttctctcaattaaactaagtcacaaactcaccattacaactCTTTGTGTGTATGTTTGTATGTTTATCA
ANX3u13	ANX3	A	G	T	T	cds	GE460	ATTCAAATGTG CTCAACTGC	CTGAGTTAAG CAAGTGCAA	ATTCAAATGTGCTCAACTGCGgttgcttttaataattttggtggtgctctcttttttagc(a/g)tttgatgaatacagaaatcacagccaaaggagacattgtgacagcataaaaaggagaattatctgggcatcttggaagacttactgttgccatagtaagacttgagctgctggttaactaagttaactTTGCACCTTGCTTTAACTCAG
ANX3u14	ANX3	T	A	I	N	cds	GE460	ATTCAAATGTG CTCAACTGC	CTGAGTTAAG CAAGTGCAA	ATTCAAATGTGCTCAACTGCGgttgcttttaataattttggtggtgctctcttttttagcatttgatgaatacagaaata(t/a)lcagccaaaggacattgtggacagcataaaaaggagaattatctgggcatcttggaagacttactgttgccatagtaagacttgagctgctggttaactaagttaactTTGCACCTTGCTTTAACTCAG
ANX3u2	ANX3	C	A	S	R	cds	GE425	CTTTTTAGGGC GCGGA	AGCAACATGG CTAAATATGTG	CTTTTTAGGGCGGGGhacaaacgaagatgcttgattgaatacttaactaccaggacaag(c/a)laggcaaatgaaggatatctctcaagcctattatacagggtgcttattttctgcttaccttcaccactgttcaCACATATTTTAGCCAAATGTTGCT
ANX3u3	ANX3	C	T	P	L	cds	GE433	TTACTTTACTAT AGATTAACCCA ATTTT	TTTCCCAGGG AATTAAGG	TTACTTTACTATAGATTTAACCCTTTCTattctccgtgaagctgaacattatttgcctttttttagcaggttaattgtgtgaggaacagc(c/t)ggccttttagcggaaagactgcagctcagaccttgaaggctggtctggaaagttcatgtgcatctctagcgtCCCTTAATTCCTTGGGAAA
ANX3u4	ANX3	A	T	E	V	cds	GE444	TCTCTATGTGT CTTTGTGACCA AT	GACTTACTTTA ATTGCTGAATA TAGG	TCTCTATGTGTCTTTGTGACCAATGacattgtgtgtgaacacctgcattctcttaacagggtatctggaactgatgagttactctgaacgaataatggtgtccagatcag(a/t)aatgaccttttgacattcgaacagagttcgaagagcattatggctattccCTATATTTCAGCAATTAAAGTAAAGTC
ANX3u5	ANX3	C	T	-	-	noncoding	GE439	TGATTCATTTA TGCTCTCCCAT T	GAACTAAGGTG GAGCTGTTGG	TGATTCATTTATGCTCTCCCATtattatactgattgttttttctcattgattttt(c/t)tttgagctcggaatactctgagagactatgaatacacactcttaaaaaactgtggtgagatgactgaacaaagaagataatctctccaaaggtccacagatgggcttttCCCAACAGCTCCACCTTACTTC
ANX3u6	ANX3	G	T	-	-	noncoding	GE448	CTTTTGTTCAA GACAAATGTTG AG	TGATCTCTTAC TGCTGTCA	CTTTTGTTCAGACAAATGTTGAGCataataatlgagtataaataactttt(g/t)tttcatttaggaactgatgagaaatgctctcagcattctgactgagaggtcaaatgcacagggcagctgattgttaaggaaataatcaagcagcagcatatggaaggtcaacattacaTGACAGGCAAGTAAAGAGATCA
ANX3u7	ANX3	A	T	E	D	cds	GE425	CTTTTTAGGGC GCGGA	AGCAACATGG CTAAATATGTG	CTTTTTAGGGCGGGGhacaaacga(a/t)gatgccttgattgaatacttaactaccaggacaagcaggcaaatgaaggatatctctcaagcctattatacagggtgcttattttctgcttaccttcaccactgttcaCACATATTTTAGCCAAATGTTGCT
ANX3u8	ANX3	T	G	-	-	noncoding	GE443	GATGTCAATTT GAACCAATG	TGTAACTGGC TGATTTGCTTC	GATGTCAATTTTGAACCAATGGactttcaagattt(g)cttctagggttggaacacaggaggaacaggttaagagattatccagacttttagcccatcagtggtgactgaagctattcagaagaatcagagggaattggtgagtgatatttacaattctcttcttaagtgtGAAGCAATTCAGGCAATGTTTACA

FIG. 5D

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
APODu5	APOD	C	A	T	K	cds	GE371	CAGCCTTCCTTG TGTGTTCCCTG	TCCTGGTTGAT TGGTTTGCT	CAGCCTTCCTTGTTGTTCTCTGagatttctcttgccctcctcccaataaatgctgcctctctctg gttcacgttatgccatcgaccgactggatcctggccaccgactatgagaactatgcccctcg tttattcctgtacctgatcatccaattttcacgtggattttgcttggatcttggcaagaac cctaactcctcctccagaaacagtggactcttaaaaaatatcctgacttctaataacattgatgt caagaaaatga c/a ggtcacagaccaggtagaaccccccaagc t/a ctcgttaaccaggttctaca gggaggctgcacccactccatgttactcttgtcttgctttccctaccacccccccccataaa GACAAACCAATCAACCACGA
APODu6	APOD	T	A	L	H	cds	GE371	CAGCCTTCCTTG TGTGTTCCCTG	TCCTGGTTGAT TGGTTTGCT	CAGCCTTCCTTGTTGTTCTCTGagatttctcttgccctcctcccaataaatgctgcctctctctg gttcacgttatgccatcgaccgactggatcctggccaccgactatgagaactatgcccctcg tttattcctgtacctgatcatccaattttcacgtggattttgcttggatcttggcaagaac cctaactcctcctccagaaacagtggactcttaaaaaatatcctgacttctaataacattgatgt caagaaaatga c/a ggtcacagaccaggtagaaccccccaagc t/a ctcgttaaccaggttctaca gggaggctgcacccactccatgttactcttgtcttgctttccctaccacccccccccataaa GACAAACCAATCAACCACGA
ARd10	AR	T	G	G	G	cds	GE655	CGGGTAAGGG AAGTAGGTG	CTTCGCTCAGG ATGCTTTTA	CGGGTAAGGGTAAGTAGGTGgaagattcagccaagctcaaggatggaagtgcagttagggctggg aagggtctacctcgccgctccaaagacctaccgaggagctttccagaattctgttccagagcg tgccgaagtgatccagaacccggcccccagccagccagagccgagcagcagcagcagcagcagc gccagtttctgtctgtcgtcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagc gcagc tctcccaagcccatcgttagagggcccccacaggtctacctgctggtgaggaacagcaaccttc acagccgagtcggccctggagtgccaccccgagagaggttgcgtcccgagagcctggagccgccc tgccgcagcagcaagggtgcgcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagc ccatccacgttgcctgtggtggcccccacttccccggcttaagcagcagcagcagcagcagcagcagc AGCATCTCTGAGCGAGG
ARd11	AR	G	C	E	Q	cds	GE655	CGGGTAAGGG AAGTAGGTG	CTTCGCTCAGG ATGCTTTTA	CGGGTAAGGGTAAGTAGGTGgaagattcagccaagctcaaggatggaagtgcagttagggctggg aagggtctacctcgccgctccaaagacctaccgaggagctttccagaattctgttccagagcg tgccgaagtgatccagaacccggcccccagccagccagagccgagcagcagcagcagcagcagcagc gccagtttctgtctgtcgtcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagc gcagc tctcccaagcccatcgttagagggcccccacaggtctacctgctggtgaggaacagcaaccttc acagccgagtcggccctggagtgccaccccgagagaggttgcgtcccgagagcctggagccgccc tgccgcagcagcaagggtgcgcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagc ccatccacgttgcctgtggtggcccccacttccccggcttaagcagcagcagcagcagcagcagcagc AGCATCTCTGAGCGAGG
ARd12	AR	G	A	E	E	cds	GE659	CCACTTTCCCC GGCTTA	CGGCCAGAGCC AGTGA	CCACTTTCCCCGGCTTAagcagctgctcgcgtgaccttaaagacatcctgagcagggccagcacc atgcaactccttcagcaacacagcagcagcagcagcagcagcagcagcagcagcagcagcagcagc gga g/a gctcgggggctcccatctctccaaaggacaattacttaggggcaactcgaccatt tctgacaacgccaagggttgtaaggcagctgcggtgctccatggccctgggtgtggagcggtt ggagcactgagtcagggaacagcttcggggggtatgcatgtacgccccacttctgggagttc caccgctgtgcgtcccatcctgtgccccatggccccatggccccatggccccatggccccatgg agcgaaggcgaagagcctaggctgctctggcagcagcagcagcagcagcagcagcagcagcagcagc tgccgtctacctgtctctctacaagtcgggagcagcagcagcagcagcagcagcagcagcagcagcagc gactactacaacttTCCACTGCTCTGCCG

FIG. 5F

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
ARd9	AR	C	T	L	L	cds	GE573	CATGCTTCCCC TC	CAGGCTGTTCT CCCTGATAAA	CATGCTTCCCTTCCCATctctgtcttcatcccaatcaggtccagtggaatgggtggaataatcaa 244 aaatctcttgatgaacttcgaatgaactacatcaaggaaact c t gatcgatcatatgcgatgca aaagaaaaaatcccaatcctgctcaagacgcttccacagctcaccagctcctggaactccgtg cagcctgaagcaaacgatggaggtgctTTATATCAGGAGAACACGCTG
ARu1	AR	G	A	K	K	cds	GE584	TTTCACTGACAT GTCTTCCAT	AGTGTCTCTCT CTGGAAGGTAA	TTTCACTGACATGTCTTCCATggttttttgggtcttctccagtttgagagactgccagggaaccatg 262 ttttggccattgactattactttccaccccaagaagaccctgctgatcttggagatgaagctctt gggtgtcactatggagcttcacatgtggaagctgcaa g a g ctcttctcaaaaagagccgtg aaggtaaagggtcttgacatgcagctctctcttcccttctctcttTACCTTCCAGAGAGAGACAC CT
ARu2	AR	T	A	L	H	cds	GE575	AAACTTCCCT CATTCCTTTT	CAAAAGTGGTC CTCTCTGAATC TC	AAACTTCCCTCATTCCTTTTctctctgtgtatctctctcccaagtlaccgcatgcacaagtcctg 244 gatgtacagccagtgtgtccgaatgaggacactctctcaagagtttgagtggtctcaaatcaacc cccaggaattctctgtgcatgaaagcactgctac t a cttcagcatltagtaagtgcctagaagt gcagggaatgccccctgagggcacagagATTTCAGAGAGGACCATCTTTG
ARu3	AR	G	C	-	-	noncoding	GE586	TCAGACTTAGC TCAACCCGTC	CCAAGCTGCTG TATTTTAGTGA G	TCAGACTTAGCTCAACCCGTCagtagtaccagactgaccactgctctgctctctctctccag 262 gcttccgcaacttacacgtggacgaccagatggctgctcatctcagtaactcctggatgggctcatg gtgttgccatgggtggcgatccttcaccaatgtcaactccaggaatgctctacttcgccccctga ctggttttcaatgagtaagt g c ctctggggccagagacCTCATAAATACAGACGCTT GG
ARu4	AR	T	A	L	-	cds	GE630	TTTTCACCACT GATGATAAATT C	AAATATGATCC CCCTTATCTCA	TTTTCACCACTGATGATAAATTCaagtctctctctctcccaatagccgggaagctgaagaaact 384 tggtaactgaaactacaggaggaaggaggtctccagaccaccagccccactgaggagacaa cccagaagctgacagtgacacattgaaggtatgaatcagccccactctcttgaaatgctctg gaagccattgagccaggtgtgagtggtgtggacacacacacacagccagccactctcttgcagc cttgctcttagctcctcaatgaaatgggagagagacagcttgcacacgtggtcagtggtggccaaag cct t a gcttggttaaggaaagggaagtgaggagcaTGAGATAGGGGGATCATATT
ARu5	AR	T	G	S	A	cds	GE1221	TTTCACTGTGTA GGATATAAATT CA	TCTGGTCTAAA GAGAGACTAGA AAAT	TTTCACTGTGTAGGATATAAATTTCATatcttctgttctagaaatccccgaagaaagagactctg 310 gaaactcattatcaggtctatcaactctgtatggtgtctccagggaaacagaagtaacctgtgc ggcagcagaaatgattgcaactatgataaattccgaaggaaatgtcca t g cttctgctgc ttcggaaatgttatgagcagggatgactctggggaggtgaagatacttctctctctctctctc ctc AAACTTCCCTCATTCCTTTTctctgtgtatctctctccaggtaccgcatgcacaagtcctcg 244 gatgtacagccagtgtgtccgaatgaggacactctctcaagatttggatggctccaaatcaacc cccaggaattctctg t g gcatgaaagcactgctactctcagcatttagtaagtgcctagaag gcagggaatgccccctgagggcacagAGATTTCAGAGAGGACCATCTTTTGG
ARu6	AR	T	G	C	G	cds	GE575	AAACTTCCCT CATTCCTTTT	CAAAAGTGGTC CTCTCTGAATC TC	AAACTTCCCTCATTCCTTTTctctgtgtatctctctccaggtaccgcatgcacaagtcctcg 616 caccatgcaactctctcagcaacagcagcagaagcagatctccgaaggcagcagcagcgaggag cgaggagccctcggggtctccactctctccaaaggacaatctactagggggactctcgaccatt tctgacaacgccaaggtgtgtgaagcagctgctgtgtccatggcctgggtgagggagcgtt ggagcatctgagtcagggggaacagcttcggggggatgtcatgtacgccccacttctggaggtc caccgctgtgctccccactctgtgccccatggccgaatgcaagggttctctctgtagacgac agcgaggcagagcactagagatactgctgagatctctctctcaaggaggttacaccaagg tgcgctctaccctgtctctctcaagtcaggagcactggcaggagcagctcgggacacttgaaac gactactacaactTCCACTGGCTCTGGCCG
ARu7	AR	T	C	S	P	cds	GE659	CCACTTTCCTCC GGCTTA	CGGCCAGAGCC AGTGGA	CCACTTTCCTCCCTTtagcagctgc t c ccgctgacttaagacatctctgagcagggccag 616 caccatgcaactctctcagcaacagcagcagaagcagatctccgaaggcagcagcagcgaggag cgaggagccctcggggtctccactctctccaaaggacaatctactagggggactctcgaccatt tctgacaacgccaaggtgtgtgaagcagctgctgtgtccatggcctgggtgagggagcgtt ggagcatctgagtcagggggaacagcttcggggggatgtcatgtacgccccacttctggaggtc caccgctgtgctccccactctgtgccccatggccgaatgcaagggttctctgtagacgac agcgaggcagagcactagagatactgctgagatctctctctcaaggaggttacaccaagg tgcgctctaccctgtctctctcaagtcaggagcactggcaggagcagctcgggacacttgaaac gactactacaactTCCACTGGCTCTGGCCG

FIG. 5G

[illegible]

FIG. 5H

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')
BDNFu3	BDNF	A	G	K	R	cds	GE184	AAGCCCTAAACC AGTTTTCG	GTTCCTCCTCT GGTCATGGA
CD36u8	CD36	G	A	P	P	cds	GE459	TGCTTTAAACA GTGACTTTGTT TTTTGT	TGCCATTCAATA TTTGCTACT
CD36u1	CD36	A	T	Q	L	cds	GE437	CTGCTGTTTCT TTAGAGTTTCG	TGTTGTTTGTTC TACTCACTGCC A
CD36u2	CD36	T	G	V	V	cds	GE437	CTGCTGTTTCT TTAGAGTTTCG	TGTTGTTTGTTC TACTCACTGCC A
CD36u3	CD36	G	T	V	F	cds	GE459	TGCTTTAAACA GTGACTTTGTT TTTTGT	TGCCATTCAATA TTTGCTACT
CD36u4	CD36	T	A	G	G	cds	GE437	CTGCTGTTTCT TTAGAGTTTCG	TGTTGTTTGTTC TACTCACTGCC A
CD36u5	CD36	G	T	V	V	cds	GE476	TGGAATGCAGC TCCTTTT	ATGGACTGTGC TACTGAGG
CD36u6	CD36	G	A	S	N	cds	GE426	TCCTAGGAATC TGCTCTATTG	CTGTGATGACC ACAAAACA
CD36u7	CD36	A	T	V	V	cds	GE440	ATCATTTGCCA CTCGATT	GCATACCTTATA CTTCAGTATCT GTCTTACC

FIG. 51

[illegible]

FIG. 5J

[illegible]

FIG. 5K

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
CYP11A u3	CYP11 A	G	A	E	K	cds	GE585	CTGCAGGGAACCTCCTCTTTTtttcttctctctccacagctgacatatacacccagaactt	CACAGGGGCA ACAAGGT	CTGCAGGGAACCTCCTCTTTTtttcttctctctccacagctgacatatacacccagaactt ctactgggaattgagacagaaaggaaggttccaccacgattaccgtggcatctctacagactcc tgggagacagcaagatgtccttc[g/a]aggacatcaaggccaacgtcacagagatgctggcagg aggggtggacacggtgaggtggtgtagggcggaacCTTGTGCCCCCTGTG	248
CYP11A u4	CYP11 A	T	A	L	Q	cds	GE556	CTACTCCCCAC CAGACG	CCAGGGATTGG AGTTGGG	CTACTCCCCAC CAGACGCTccatgacctgacgtggcacttgtatgagatggcacgcaacctgaag gtgcaggatagtgtcgggcagaggtcttggctgcggcgccacaggccaggagagacatggccac gatgtacagctggtccccctctcaagccagcatcaaggagacac[t/a]aaggcaagccccac aaccacccatgccgccccCAACTCCAATCCCTGG	229
CYP11B 1a30	CYP11 B1	A	T	N	I	cds	GE570	TCCCAGCACCA AAGTCTGAG	GGCATCACCT CTCTGGGT	TCCCAGCACCA AAGTCTGAGggtctgctcccgctccccggtataggcgacaactgtatccagaaaa tctatcaggaaactggccttcggccgctccacagatcacaccagcatcgtggcgagctcctgttg a[a/t]tgcggaactgtgcagatgccatcaaggccaactctatggaaactactgcaggagcg tggacacggtcagggcggaacccagccccACCAGAGAGGGTGTGATGCC	243
CYP11B 1a31	CYP11 B1	A	G	-	-	noncoding	GE577	GAATGGCCTG AATGGC	CTCCAGGGTCT CTGAGGCTG	GAATGGCCTG AATGGCgcttcaaccgattgcggctgaatccagaaagtgtgtcgcccaacgctg tgcagaggttctctcccgatgggtgagatgcagtgccagggaacttctccaggccctgaagaaag gtgtgcagaaacggcggggagcctgacctggagctccagccagcatcttccactacacal agaaggtgtggggcac[a/g]tgggaagatccAGGCTCAGAGACCTCGAG	246
CYP11B 1a32	CYP11 B1	A	T	-	-	noncoding	GE617	ATGGCACTCAG GGCNAA	AGGGCTCTGGG TGTGCC	ATGGCACTCAG GGCNAAaggcagaggtgtgcattggcagtgccctggtctccctgcaaaagjrcaca ggcacttgggcaacagagcgccgggtccccaggagagctgtgccccttgaagccatgccccagc gtccaggcaacaggtggctgagctgagctgcagatctggaggagagcagggttatgaggacctgcac ctggaagtacacccagaccttccaggaactggggcccatcttcaggtaagacctcctctggccc[a /t]cgctGGGAACACCCAGAGCCCT	285
CYP11B 1a33	CYP11 B1	C	T	-	-	noncoding	GE625	GGAGGCAGCCA GGAGGC	GTGTCCCTTCC CCATAGCAC	GGAGGCAGCCA GGAGGC[t/t]cggggtgcttgtgtcagcagtgcatctccccgaagccag caacttggctcttcttggagagcggtggcgctggttggccacagcccaagttctgcccagctg aacttctccatgcccctggaggtcatgttcaaatccacgtccagctccatgttcatgcccaggag cctgtctcgtggaccagcccccaagggtgtggaaggagcacttggggcctgggactgcatcttcc agtaacggtgagggccagggacccggggaGTGTATGGGGAAGGACAC	307
CYP11B 1d24	CYP11 B1	G	C	-	-	noncoding	GE1231	GCAACTTTGAG GGTCTGAGAA	CCTGGGTTCAG GCAGAAA	GCAACTTTGAG GGTCTGAGaggtgcacacgtcgtatgggtgcggaccagcagatggaac cc[g/c]gctgtgcacacaggtgtggaacacccctccaggtggagacactaacccaaaggagacata aagatgggtcacagcttcattatggggccagcatgttccccctcctcactcagagccatcaa ctaatacagctctctgcacccagggtccccagcctggccacccagcctctcttTCTGCTGACCCCTAG G	261
CYP11B 1d25	CYP11 B1	T	C	Y	Y	cds	GE570	TCCCAGCACCA AAGTCTGAG	GGCATCACCT CTCTGGGT	TCCCAGCACCA AAGTCTGAGggtgctcccgctccccggtataggcgacaactgtatccagaaaa tcta[t/c]caggaaactggccttcagccgctcccaacagtaacacagatcgtggcgagctcct gttgaatgcggaactgtccagatgacataaggccaactctatggaaactactgcaaggagcg tggacacggtcagggccggcaacccagccccACCAGAGAGGGGTGATGCC	243
CYP11B 1d26	CYP11 B1	C	G	N	K	cds	GE577	GAATGGCCTG AATGGC	CTCCAGGGTCT CTGAGGCTG	GAATGGCCTG AATGGCgcttcaaccgattgcggctgaatccagaaagtgtgtcgcccaa[c/g] gctgtgcagaggttctctcccgatgggtgagatggcagggagcttctccaggccctgaagaa gaaggtgtgcagaaacggccggggagcctgacctggagctgacgttccagccagcatcttccactaca ccatagaaggtgtggggccacatggggaagatccAGGCTCAGAGACCTCGAG	246

FIG. 5L

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
CYP11B 1d27	CYP11 B1	C	A	-	-	noncoding	GE582	CTCCTGTGCAAGGCTG	CTCCAGCAGGG GGCCAG	CTCCTGTGCAAGGCTGacccctgagctgtgtctctctgcagacgggtgttctctctgctgatgaac gctctttgagctggctcggaaccccaacgtgcagcagggccctgcgcagagagcctggccgccc gaggccagcatcagtgaaacatccccagaaggaacacccagctnccccctgtctgctgctgcccct caaggagaccttg c/a gggggtgctggtgagggcctccctgtggccctggcccttgcctgctgga G	261
CYP11B 1d28	CYP11 B1	C	T	-	-	noncoding	GE617	ATGGCACCTCAG GGCAAA	AGGGCTCTGGG TGTTCCT	ATGGCACCTCAGGGCAAAggcagaggtgtgcatggcagtgccctggctgtccctgcaaaagggcaca ggcactgggcacgagagcggcccggtcccccagacagtgctgccccttgaagccatgccccagc gtccaggcacaaggtggctgagggctgctgcagatctggaggagcaggggttatgaggacctgcac ctggaaagtacacacagaccttccaggaaactggggcccattttcaggtaaaagccctcc c/l lygc ccacgctGGGAACACCCAGAGCCCT	285
CYP11B 1d29	CYP11 B1	C	T	-	-	noncoding	GE625	GGAGGCAGCCA GGAGGC	GTGTCCCTTCC CCATAGCAC	GGAGGCAGGCAGGAGGCC c/l ggggctgcccctgtgctcagcagtgcatctccccgaaagccag caacttggtctcttttggagagcggctggggcctgggtggccacagcccccaagttctgcccagcctg aacttctccatgcccctggaggtcatgttcaaatccacccgctccagctcatgttcatgcccagtag cctgtctgctggaccagccccaaaggtgtggaaagagcactttgagggcctgggactgcatcttcc agtacgggtgagggccaggggacccgggcaGTGCTATGGGAAGGGACAC	307
CYP11B 1u1	CYP11 B1	A	G	Q	R	cds	GE617	ATGGCACCTCAG GGCAAA	AGGGCTCTGGG TGTTCCT	ATGGCACCTCAGGGCAAAggcagaggtgtgcatggcagtgccctggctgtccctgcaaaagggcaca ggcactgggcacgagagcggcccggtcccccagacagtgctgccccttgaagccatgcccc a/ g gcgtccaggcaacaggtggctgaggtgctgcagatctggaggagcaggg tatgaggacct gcaactggaaagtacacacagaccttccaggaaactggggcccattttcaggtaaaagccctccccggc ccacgctGGGAACACCCAGAGCCCT	285
CYP11B 1u10	CYP11 B1	C	G	P	A	cds	GE582	CTCCTGTGCAAG GGTCTG	CTCCAGCAGGG GGCCAG	CTCCTGTGCAAGGTCTGacccctgcagctgtgtctctctgcagacgggtgttctcccttgcctgatgaac gctctttgagctggctcggaaccccaacgtgcagcagggccctgcgcagagagagcctggccgccc gaggccagcatcagtgaaacatccccagaaggaacacccagagctn c/g cccttgcctgctgctgg cctcaaggagaccttgcgggtgctggctgagggcctccctgtggccctGGCCCCCTGCTGGA G	261
CYP11B 1u11	CYP11 B1	T	G	-	-	noncoding	GE582	CTCCTGTGCAAG GGTCTG	CTCCAGCAGGG GGCCAG	CTCCTGTGCAAGGTCTGacccctgcagctgtgtctctctgcagacgggtgttctcccttgcctgatgaac gctctttgagctggctcggaaccccaacgtgcagcagggccctgcgcagagagagcctggccgccc gaggccagcatcagtgaaacatccccagaaggaacacccagagctnccccctgtctgctg g gcgg cctcaaggagaccttgcgggtgctggctgagggcctccctgtggccctGGCCCCCTGCTGGA G	261
CYP11B 1u12	CYP11 B1	C	T	L	L	cds	GE531	ACAGGAAGCCC CATCCA	AGGTCTCTCAGC TCGAGGGGT	ACAGGAAGCCCCATCCAGctgaggaaccccttctctatggalgcceccacccctccaggctctaccctgt gggctctgttt c/t tgagcaggtggctgagctcagacttgggtcttcagaactaccacatccca gctgggtgagtgagcggccacACCCCTCGAGCTGAGAACCT	171
CYP11B 1u13	CYP11 B1	T	C	L	L	cds	GE618	CTCCCCAGTCA TTCCCTGA	GGCCATGCTGC CCAGAC	CTCCCCAGTCAATTCCTGATccccgctctgcacccgtccgcagacatlggtgcggtgttccctcta ctctctgggtgcgaaccccgctgttcccagggcctgagcgtataaaccacccagcgtggctag acatcagggggtccggcaggaacttctaccagtgcccttggcttggcatggcagcagtgctt t/c gggcggcgttggcagagggcagagatgctgctgctgcgcacacatgtgagcagggccccgggc tggggagggggcctgggcgggtGTGGGACGATGGGC	297
CYP11B 1u14	CYP11 B1	G	A	C	Y	cds	GE617	ATGGCACCTCAG GGCAAA	AGGGCTCTGGG TGTTCCT	ATGGCACCTCAGGGCAAAggcagaggtgt g/a catggcagtgccctggctgtccctgcaaaaggg cacaggcactgggcacgagagccgggtccccagggagagtgctgccccttgaagccatgccc cagcgtccaggcaacaggtggctgaggtgcgtgcagatctggaggagcaggggttatgaggacct gcaactggaaagtacacacagaccttccaggaaactggggcccattttcaggtaaaagccctcc tggc ccacgctGGGAACACCCAGAGCCCT	285

FIG. 5M

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
CYP11B lu15	CYP11 B1	G	A	R	H	cds	GE617	ATGGCACTCAG GGCAA	AGGGCTCTGGG TGTTC	ATGGCACTCAGGGCAAAggcagagggtgctgcattggcagtgccclggctgctccctgcaaaaggacaca ggcactgggcacgagagcgccgggtcccaggaagtcctcccttgaagccatgccccagc (g/a)ccaggcaaacagggtggctgaggtgctgcagatctggaggagcaggggttatgaggacct gcacctggaagtacaccagaccttccaggaaactggggccccattttcaggtaaaagccctccctggc ccacgctGGGAACACCACCCAGAGCCCT
CYP11D lu16	CYP11 B1	C	T	D	D	cds	GE536	ATGTTCCCATT CCAGCAC	AGCAAGAACAC GCCACA	ATGTTCCCATTCCAGCACggcctcgctgctggccccacacaggtacga c/t tgggaggagcagg catggtgtgtgatgctgcgggagagctggagaagctgcaacaggtggaacagctgcaccc acaggatgagcctggagccctgggtggcctacagacaacatcgtyggcacaaaTGTTGGCGTGTTC TTGCT
CYP11B lu17	CYP11 B1	G	A	L	L	cds	GE625	GGAGGCAGCCA GGAGGC	GTGTCCCTTCC CCATAGCAC	GGAGGCAGCCAGGAGGccgggctgcttggctcagcagtgcatctctccccccaagrcagcaac ttlg/a g ctctttttggagagcggtggcctgggtggccacagcccccaagltctggcagctg aatctctccatgctcctggaggtcatgtcaaatccacgtccagctcatgttcatgcccaggag cctgtctcgctggaccagccccaaagggtgtggaaggagcaactttgaggcctgggaactgcatactcc agtacggtgaggccaggacccgggcaGTCTATGGGAAGGGACAC
CYP11B lu18	CYP11 B1	C	T	-	-	noncoding	GE570	TCCAGCACCA AAGTCTGAG	GGCATCACCTT CTCTGGGT	TCCAGCACCAAAGTCTGAGggctgctcc c/t gctccccgataggcgacaactgtlatccag aaaaatctatcaggaactggcctcagccgcctcaacaglacaccagcatcgtggcgagctcct gttgaatcggaactgtcccgagatgccatcaaggccaactctatggaactcactgcaggaggcg tggaacagggtcaggccggaaccagccccACCAGAGAGGGGTGATGCC
CYP11B lu19	CYP11 B1	G	A	A	T	cds	GE582	CTCCTGTGCAA GGTCTG	CTCCAGCAGGG GGCCAG	CTCCTGTGCAAGGTCTGacctgcagctgctctctcctgcagacgggtgtttcccttgtgatgaac gctctttgagctggctgggaacccccaaagtg c agcagcctgcgcagagagcctggccgccc g/a cagcagcatcagtgaacatccccagagggcaacacccagcagctncccttgcctgclgagg ccctcaaggagaccttgcgggtgggtgctggctgaggcctccctgtggcctTGGCCCCCTGCTGGA G
CYP11B lu2	CYP11 B1	A	G	E	E	cds	GE617	ATGGCACTCAG GGCAA	AGGGCTCTGGG TGTTC	ATGGCACTCAGGGCAAAggcagagggtgctgcattggcagtgccctggctgctccctgcaaaaggacaca ggcactgggcacgagagcgccgggtcccaggaagtcctcccttgaagccatgccccagc gtccaggcaacagggtggctgaggtgctgcagatctggaggagcaggggttatgaggacctgcac ctggaagtacaccagaccttccaggga a/g ctggggccccattttcaggtaaaagccctccctggc ccacgctGGGAACACCACCCAGAGCCCT
CYP11B lu20	CYP11 B1	T	C	V	A	cds	GE531	ACAGGAAGCCC CATCCA	AGGTCTCAGC TCGAGGGGT	ACAGGAAGCCCCATCCAgctgaggacccttctatgtagtccccccacctccaggtctctaccctgt gggtctgttctggagcagtg t/c gagctcagacttggtgcttcagaactaccacatccca gctgggtgagtgagcggccacACCCCTCGACTGAGAACCT
CYP11B lu21	CYP11 B1	C	G	R	R	cds	GE618	CTCCCCAGTCA TTCCCTGA	GCCCATGCTGC CCAGAC	CTCCCCAGTCATTCCTGAtccccgctctgcaccgtcgcagacatatgggtgcgclgtctctcta ctctctgggtcgcaacccgcttgttcccgagggcctgagcgtataacccccagcgtgggtag acatcaggggtctccgcaggaacttctaccagtgcccttggccttggcatggcagtgccctt gggcggc c/g ctgcagagggcagagatgctgctgctgcaccatgtgagcagggccccggc tggggaggggcctgggggGTCTGGGCAGCATGGGC
CYP11B lu22	CYP11 B1	A	T	T	S	cds	GE625	GGAGGCAGCCA GGAGGC	GTGTCCCTTCC CCATAGCAC	GGAGGCAGCCAGGAGGccgggctgcttgtgctcagcagtgcatctctccccccaagrcagcaac ttggctcttttggagagcggtggcctgggtggccacagcccccaagltctgcccagcctgaact tctccatgctcctggaggtcatgttcaatccacgtccagctcatgttcatgcccaggagcctg tctcgctgg a/t ccagccccaaagggtgtggaaggagcaactttgaggcctgggaactgcatactcc adtacgtgaagccaggaacccggcagTGTATGGGAAGGGACAC

FIG. 5N

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')
CYP11B lu23	CYP11B1	C	G	-	-	noncoding	GE577	GAATGGGCTG AATGGC	CTCCAGGGTCT CTGAGGCTG
CYP11B lu3	CYP11B1	G	A	L	L	cds	GE617	ATGCCACTCAG GGCAAA	AGGCCTCTGGG TGTTCCC
CYP11B lu4	CYP11B1	C	A	F	L	cds	GE577	GAATGGGCTG AATGGC	CTCCAGGGTCT CTGAGGCTG
CYP11B lu5	CYP11B1	A	G	K	R	cds	GE577	GAATGGGCTG AATGGC	CTCCAGGGTCT CTGAGGCTG
CYP11B lu6	CYP11B1	C	T	T	I	cds	GE625	GGAGCGAGCCA GGAGGC	GTGTCCCTTCC CCATAGCAC
CYP11B lu7	CYP11B1	G	A	A	A	cds	GE570	TCCACGACCA AAGTCTGAG	GGCATCACCT CTCTGGGT
CYP11B lu8	CYP11B1	G	A	-	-	noncoding	GE570	TCCACGACCA AAGTCTGAG	GGCATCACCT CTCTGGGT
CYP11B lu9	CYP11B1	A	G	N	D	cds	GE582	CTCCTGTGCAA GGTCTG	CTCCAGCAGGG GGCCAG
CYP11B 2d22	CYP11B2	C	T	-	-	noncoding	GE1213	GAGGACTGAAG GGAGTGTG	CCACTGGGTGG TGGAGA

FIG. 50

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Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
CYP11B 2d23	CYP11B2	G	A	-	-	noncoding	GE533	CATCCAGCTGA GGACCCCTT	ACTGGGAGGG AGGTTCTC	CATCCAGCTGAGGACCTTTct [g/a] tggatgcccccaactccaggctctaccctgtgggtctg cttttggagcagtggtgagctcagacttgggtgcttcagaactaccacatcccagctgggtgag tgagccccacacccctcgagctGAGAACCTCCCTCCCACT	172
CYP11B 2d24	CYP11B2	A	G	N	S	cds	GE587	ATGCTTCCAG CACCAAGAT	GGCATCACCT CTCTGGG	ATGCTTCCAGCACCAAGATctgagggtgtccccctgtccctggacaggtgacaactgtatcca gaaaatctaccaggaactggccctca [a/g] ccgcccccaacacacacacaggtcatcgtggcagag ctccctgtgaaaggcgaactgtcactagaagccatcaaggccaactctatggaaactcactgcagg gagcgtggaacacggtcaggccagcaaccagcccccacCCAGAGAGGGTGTATGCC	248
CYP11B 2d25	CYP11B2	A	C	R	R	cds	GE588	GAGTCTCTCTG TGCAAGGTC	CTCCAGCAGGG GGCCAG	GAGTCTCTCTGTCAGGTCagacctgcagacatggctctctgtagacagcttccccctgtga tgacgctctttgagctggctcggaaccccgacgtgcagcagatctctgcgacagagagcctggcc gccgacccagcatcagtgaaacatccccagaaagcaaccacagctgccctgtctgctggcgcc cctcaaggagaccttg [a/c] ggtgggtgctggatgaggcctccctgtggcctGGCCCCCTGCT GGAG	264
CYP11B 2d26	CYP11B2	G	A	-	-	noncoding	GE610	CCTGTCTCTTG CTGGG	CAGGGTCTCTG GGGCTG	CCTGTCTCTGCTGGGGggccctcacaaagctctgccctggcctctgtaggaaatggcctgaatgg cgcttcaaccgatlgcggctgaacccagatgtgctgtgcgcccaggcgtgcagaggttccctccc gatgggtgagtgagtgccagggacttctccagggcctgaagaagaaggtgctgcagaacgccc gggggagcctgacctggagctccagcccgacatcttccactacacacatagaaggtgtgggccc gcgggaag [g/a] ccAGGCCCCAGAGACCCCTG	292
CYP11B 2u1	CYP11B2	A	G	K	R	cds	GE610	CCTGTCTCTTG CTGGG	CAGGGTCTCTG GGGCTG	CCTGTCTCTGCTGGGGggccctcacaaagctctgccctggcctctgtaggaaatggcctgaatgg cgcttcaaccgatlgcggctgaacccagatgtgctgtgcgcccaggcgtgcagaggttccctccc gatgggtgagtgagtgccagggacttctccagggcctga [a/g] gaagaaggtgctgcagaac gcccgggggagcctgacctggagctccagcccgacatcttccactacacacatagaaggtgtggg ccatgcgggaaggtccAGGCCCCAGAGACCCCTG	292
CYP11B 2u10	CYP11B2	G	T	R	R	cds	GE588	GAGTCTCTCTG TGCAAGGTC	CTCCAGCAGGG GGCCAG	GAGTCTCTCTGTCAGGTCagacctgcagacatggctctctgtagacagcttccccctgtga tgacgctctttgagctggctcggaaccccgacgtgcagcagatctctgcgacagagcctggcc gccgacccagcatcagtgaaacatccccagaaagcaaccacagctgccctgtctgctg [g/t] g cgccctcaaggagaccttgagg [g] ggtgctggatgaggcctccctgtggcctGGCCCCCTGCT GGAG	264
CYP11B 2u11	CYP11B2	G	A	A	T	cds	GE637	TCCTGGCTGAG ATAAAGGATT T	AGGATCTGGG TGTTCCC	TCCTGGCTGAGATAAAGGATTtgggtgaacaggttgaggagcattggaatggcactcaggg caaaagcagaggtgtgctggcagcgcctgtgctccctgcaaaaggcagcggcactgggcact agagcc [g/a] tctgggccccctaggacgtgtgctcccttgaagccatgccccagcactccaggca acaggtggctgaggtgtgctgagatctggaggagcaggttatgagcacctgcacctggagatg caccagaccttccaggagctggggcccatttccaggtaaagccccctccctggcctcgtGGGAAC ACCCAGATCCCT	337
CYP11B 2u12	CYP11B2	G	A	P	P	cds	GE637	TCCTGGCTGAG ATAAAGGATT T	AGGATCTGGG TGTTCCC	TCCTGGCTGAGATAAAGGATTtgggtgaacaggttgaggagcattggaatggcactcaggg caaaagcagaggtgtgctggcagcgcctgtgctccctgcaaaaggcagcggcactgggcact agagcc [g/a] tctgggccccctaggacgtgtgctcccttgaagccatgccccagcactccaggca acaggtggctgaggtgtgctgagatctggaggagcaggttatgagcacctgcacctggagatg caccagaccttccaggagctggggcccatttccaggtaaagccccctccctggcctcgtGGGAAC ACCCAGATCCCT	337
CYP11B 2u13	CYP11B2	A	G	A	A	cds	GE587	ATGCTTCCAG CACCAAGAT	GGCATCACCT CTCTGGG	ATGCTTCCAGCACCAAGATctgagggtgtccccctgtccctggacaggtgacaactgtatcca gaaaatctaccaggaactggcctcaaccgcccccaacacacacacaggtcatcgtggc [a/g] gag ctccctgttgaaggcgaactgtcactagaagccatcaaggccaactctatggaaactcactgcagg gagcgtggaacacggtcaggccagcaaccagcccccacCCAGAGAGGGTGTATGCC	248

FIG. 5P

FIG. 5B

FIG. 5S

[illegible]

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer [5' -> 3']	Reverse Primer [5' -> 3']	Assay Sequence
CYP21d 27	CYP21	C	T	P	L	cds	GE612	CTCTTTGAGCTA TAAGTGGCACC	CCTCCTATGGT GAGGGC	CTCTTTGAGCTATAAGTGGCACCCTcaggggccctgacggggcgctcgtcgccatgctgctcctgggcccgtg ctgctgctgctgcccctgctggtggcgcccgctgctggaactggtggaactcccgagcctcccgagcct ccacctcc[c/t]gctcttgccegggcttcttgcaactgctgagcccgacccctcccaatctat ctgctggcctgactcagaaaattcgggcccatctacaggctccaccttgggctgcaagggtgagag gctgactcgtctcgccctCACCATAGGAGG
CYP21d 28	CYP21	A	C	P	P	cds	GE612	CTCTTTGAGCTA TAAGTGGCACC	CCTCCTATGGT GAGGGC	CTCTTTGAGCTATAAGTGGCACCCTcaggggccctgacggggcgctcgtcgccatgctgctcctgggcccgtg ctgctgctgctgcccctgctggtggcgcccgctgctggaactggtggaactcccgagcctcccgagcct ccacctcccgcccttggcccggtcttcttgcaactgctgagcccgacccctcc[c/t]atctat ctgcttggcctgactcagaaaattcgggcccatctacaggctccaccttgggctgcaagggtgagag gctgactcgtctcgccctCACCATAGGAGG
CYP21d 29	CYP21	C	T	-	-	noncoding	GE629	TGTTGCCACTC TGTACTCCTCT C	GTTGCTGGGAA GGAGCC	TGTTGCCACTCTGTACTCTCTTCCcaggccagcgctcagcc[c/t]gctccttaccctctg caggagacctgctggcaggccagtgaggagacatgagactacatgctccaagggtggcgca ggcagcatggaagggtctgtgacagctctcggaaggcagctgacacatgctgagtgagcc tcttgatcggtggcactgagaccacagcaaacacctctctctggcggtggttttttggcttcac cacctgaggtgctgctgggacacagcaaaGGCTCTTCCACGCAAC
CYP21u 1	CYP21	T	C	L	L	cds	GE612	CTCTTTGAGCTA TAAGTGGCACC	CCTCCTATGGT GAGGGC	CTCTTTGAGCTATAAGTGGCACCCTcaggggccctgacggggcgctcgtcgccatgctgctcctgggcccgtg ctgctgctgctgcccctgctggtggcgcccgctgctggaactggtggaactcccgagcct ccacctcccgcccttgccegggtcttcttgcaactc[t/c]tgcgagcccgacccctcccaatctat ctgcttggcctgactcagaaaattcgggcccatctacaggctccaccttgggctgcaagggtgagag gctgactcgtctcgccctCACCATAGGAGG
CYP21u 10	CYP21	C	T	P	L	cds	GE629	TGTTGCCACTC TGTACTCCTCT C	GTTGCTGGGAA GGAGCC	TGTTGCCACTCTGTACTCTCTTCCcaggccagcgctcagcccgctccttaccctctg agagctcgtggcaggccagtgaggagacatgagactacatgctccaagggtggcgagc[c /t]gagcatggaagggtctgtgacagctctcggaaggcagctgacacatgctgagtgagcc tctgacgggtggcactgagaccacagcaaacacctctctctggcggtggttttttggcttcac cacctgaggtgctgctgggacacagcaaaGGCTCTTCCACGCAAC
CYP21u 11	CYP21	C	T	S	S	cds	GE544	CCGAGCCACAGG GAGGTC	AGCTCCACCA CATTTT	CCGAGCCACAGGAGGTCTctggccagctctaaactcagcccccttcagcatctc[t]ggctac gacatccctgagggcacagtcatctccgaacctccaggcgccccctggatgagacggctg ggagaggccacatgagttctggcctgglatgtggggggccgggggcttgccgtgAAATGTGTGTG GAGGCT
CYP21u 12	CYP21	C	G	H	Q	cds	GE544	CCGAGCCACAGG GAGGTC	AGCTCCACCA CATTTT	CCGAGCCACAGGAGGTCTctggccagctctaaactcagcccccttcagcatctcgggctacgaca tccctgagggcacagtcatctccgaacctccaggcgcccc[c/t]gctggatgagacggctg ggagaggccacatgagttctggcctgglatgtggggggccgggggcttgccgtgAAATGTGTGTG GAGGCT
CYP21u 13	CYP21	G	A	A	T	cds	GE612	CTCTTTGAGCTA TAAGTGGCACC	CCTCCTATGGT GAGGGC	CTCTTTGAGCTATAAGTGGCACCCTcaggggccctgacggggcgctcgtcgccatgctgctcctgggcccgtg ctgctgctgctgcccctgctggtggc[tg/a]lcccgctgctggtggaactggtggaactccgga gctccacctcccgcccttgccegggtcttctgcaactgctgagcccgacccctcccaatctat ctgcttggcctgactcagaaaattcgggcccatctacaggctccaccttgggctgcaagggtgagag gctgactcgtctcgccctCACCATAGGAGG
CYP21u 14	CYP21	C	T	-	-	noncoding	GE612	CTCTTTGAGCTA TAAGTGGCACC	CCTCCTATGGT GAGGGC	CTCTTTGAGCTATAAGTGGCACCCTcaggggccctgacggggcgctcgtcgccatgctgctcctgggcccgtg cctgctgctgctgctgcccctgctggtggcgcccgctgctggtggaactggtggaactccgga gctccacctcccgcccttgccegggtcttctgcaactgctgagcccgacccctcccaatctat ctgcttggcctgactcagaaaattcgggcccatctacaggctccaccttgggctgcaagggtgagag gctgactcgtctcgccctCACCATAGGAGG

FIG. 5U

[illegible]

FIG. 5V

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
DRD1d1	DRD1	A	G	.	-	noncoding	GE1173	CTGACCCCTAT TCCCTGCTT	CTCTCCAAGGC CGCAATG	CTGACCCCTATTCCTGCTT a/g ggaacttgaggggtgtcagagccctgatgtgctttcttct taggaaagatgaggaactctgaacacctctgccatggacgggactgggctgggtgylgnagaggggact tctctgttcgtatccctcaactgctgttctctgctgtctcatctgtccacgctcctctgggaac agctggctctgtgctgccgtatcaggttcgcacacctgcgggtccaaggtgacccaacttcttgt catctccttggtgtgctcagatctcttggtggcgtctctggtcatgccctggaagcagtggtg agattgctggctctggccttgggtctctctgttaacatctgggtggccttggacatcatgtgc tccactgcatccatctcaacctctgtgatcagcgtggacaggtattgggtatctccagccc ttccgggtatgagagaagatgagcccccaaggcagccttcatctgacagtgagtggtgacct tgtctgtactcatctcttcatccagtgagctcagctgagcagctgacacaggaacacacagccc tctgatggaaatgccacttccctggctgagaccatagacaaactgactccagctcagcaggac atatgccatctcatctctgttaagcttttaccatccctgtggcctcatgtgtcacctaca ccaggatctacaggattgctcagaacaataacggcgCATTCGGCCTTGGAGAG
DRD1u1	DRD1	T	C	F	F	cds	GE1173	CTGACCCCTAT TCCCTGCTT	CTCTCCAAGGC CGCAATG	CTGACCCCTATTCCTGCTT Taggaacttgaggggtgtcagagccctgatgtgctttctttagg aagatgaggactctgaacacctctgccatggacgggactgggctgggtggtgagagggacttctc tgtctgtatccctcaactgctgttctctgctgtctcatctgtccacgctcctctgggaacacgc tggctgtgtgctggcttatacaggttcagacacctgcgggtccaaggtgacccaacttcttgcac tcttggctgtgctcagatctcttggtggcgtctctggtcatgccctggaagcagtggtgctgagat tgtggcttctggccttgggtctctctgtaacatctgggtggcctt t/c gacatcatgtgc tccactgcatccatctcaacctctgtgtgatcagcgtggacaggtattgggtatctccagccc ttccgggtatgagagaagatgagcccccaaggcagccttcatctgacagtgagtggtgacct tgtctgtactcatctcttcatccagtgagctcagctgagcagctggaacacacacagccc tctgatggaaatgccacttccctggctgagaccatagacaaactgactccagctcagcaggac atatgccatctcatctctgttaagcttttaccatccctgtggcctcatgtgtcacctaca ccaggatctacaggattgctcagaacaataacggcgCATTCGGCCTTGGAGAG
DRD1u10	DRD1	G	C	G	R	cds	GE1170	GGATCTACAGG ATTGCTCAGAA AC	GCATCTCCTC TAGCTTTTGG	GGATCTACAGGATTGCTCAGAA Caataacggcgcatctgcggccttggaagggcagagtcac gcaagaattggcagaccaccacaggtaaatggaagcctgtcgaatgttctcaacgggaaagtcc tttaagatgtccttcaaaagagaactaaagtctgaagactctgcggtgatcatgggtgtgt ttgtgtgctgttggctaccttctcatcttgacttgacttctggccttctgtggtctggggag acgcagccctctgcatgtattccaacaccttgacgtgttgggtgggttgggtgggttaattc atccttgaaccccatcattatgctttaaagctgatttctcgaagggcatttcaacccctcttag gatgctacagacttgccttgacgacgaataatgccatagagacgtgagatcaataacaatggg gcgcgatgtttccagccatcatgagccacga g/c gctccatctccaaggagtgcaatctgg tttacctgatccacatgctgtgggtcctctgaggaacctgaaaaggaggaggcagctggcatc ggcagacccttggaagagctgtcccagccctatcggtcatattggactatgacactgacgtctc tctggagaagatccaacccatcacacaacacggctcagcaccacacacacacacacacacac tgccacacatgctcatcCCAAACCTAGAGGAGATTGC

FIG. 5X

FIG. 5Y

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
DRD1u5	DRD1	A	C	A	A	cds	GE1173	CTGACCCCTAT TCCCTGCTT	CTCTCCAAGGC CGCAATG	CTGACCCCTATTCCCTGCTTAGgaacttgaggggtgcagagccctgatgctctctcttagg aagatgaggactctgaacacctctgccatggacgggactgggctgggtggagaggactctctc tgctgtatctctactgctgttctctgtcgtctcatcctgtccacgctcctctggggaacagc tggtctgtgctgcegttatcaggttcgcacacctgggtcccaagtgaccaaactctctgtctatc tccttggctgtgtcagatctcttgggtggcgtcctgtgtcatgacctggaaggcagtggtgagat tgctggcttctggcccttgggtcctctgttaacatctgggtggccttgcacatcatgtgtctcca ctgc[a/c]tccatcctcaacctctgtgtgatcagcgtggacaggtattgggtctatctccagccc tttccggtatgagagaagatgaccccaaggcagcctctcatcctgacgtgtggtgacgtgacct tgctgtactcatctctctatccacgtgcagctcagctggcacaaaggcaaaccccaaggcccc tctgatggaatgccacttccctggctgagaccatagacaactgtgactccagcctcagcaggac atatgccatctcatctctgtaatgaagctttacatctctctggccatcatgattgtcacctaca ccagatctacaggattgctcagaacaaatacggcgCATTTGGGCTTGGAGAG
DRD1u6	DRD1	A	G	K	E	cds	GE1170	GGATCTACAGG ATTGCTACAGAA AC	GCAATCTCTTC TAGCTTTTGG	GGATCTACAGGATTGCTCAGAAACaaatcggcgcatctcggccttgagagggcagcagctccac gccaagaattgccagaccacacaggttaattggaagacctgtcgaaatgtctctcaaccggaaagtctc ttttaagatgtctctcaaaagagaactaaagctctgaagactctgtcggtgatcatgggtgtgtg ttgtgtgctgttggctacaccttctctcatcttgaaactgactttgacctctgtgggtctggggag acgcagccctctgcatgtattccaacaccttgaagctgttctgtgtgttgggtgggttaattc atcctgaaccccatcttatgctcttaagtctgatttctggaaggcatttcaacctcttag gatgtcacagacttggcctcggaacaaatgccaatagagacggtgagatcaataacaatggg ggcgcatgttttccagccatcatggccacagggctccatctccaaaggagtgaactctgggttta cctgatccacatgctgtgggtctctctgagacctgaaagaggagggagcagctggcctcgcca gaccttggag[a/g]agctgtccacgacctatcggtctatattgacctatgacactgacgtctc tctggagaagatccaaccccatcacaaaacggctcagaccccaacctgaactcgcagatgaatccc tgccacacatgctcatCCCAAGCTAGAGGAGATTGG
DRD1u7	DRD1	A	C	R	R	cds	GE1173	CTGACCCCTAT TCCCTGCTT	CTCTCCAAGGC CGCAATG	CTGACCCCTATTCCCTGCTTAGgaacttgaggggtgcagagccctgatgctctctcttagg aagatgaggactctgaacacctctgccatggacgggactgggctgggtggagaggactctctc tggtgtatctctactgctgttctctgtcgtctcatcctgtccacgctcctctggggaacagc tggtgtgtgctgcegttatcaggttccg[a/c]cacttgcgggtcccaagtgaccaactctcttgt catctcttggctgtgtcagatctcttgggtggcgtcctgggtcatgacctggagggcagtggtctg agattgctggcttctggcccttgggtcctctgtgaacatctgggtggccttctgacatcatgtgc tccactgcatccatctctcaacctctgtgtgatcagctggacaggtattgggtctatctccagccc tttccggtatgagagaagatgaccccaaggcagcctcatcctgatcaggtgtggcagacct tgctgtactcatctctcatcccgatgcagctcagctggcacaaaggcaaaccccaaggcccc tctgatggaatgccacttccctggctgagaccatagacaactgtgactccagcctcagcaggac atatgccatctcatctctgtaatgaagctttacatctcttggccatcatgattgtcacctaca ccagatctacaggattgctcagaacaaatacggcgCATTTGGGCTTGGAGAG

FIG. 5Z

[illegible]

FIG. 5AA

FIG. 5BB

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
DRD2u7	DRD2	C	G	-	-	noncoding	GE1315	GCACCTGGGTGTGGTGTGTT	GGTCTCCAGGACTGAAGTT	GCACCTGGGTGTGGGTGTTTCCAGCGTGCCTCCCGGCTCTGGGACCAAGCCTGACCATGCCCTCTCCCGAGGCGTGTTCATCATCTGCTGGCTGCCCTTCTCATCACACATCTCTGAACATACACTGTGACTGCAACATCCCGCTGTCTGTACAGCGCTTCACTGCTGGCTGTATGTCAACAGCGCCGTGAACCCCATCATCTACACCACTTCAACATTTAGTTCGCAAGGCTTCTTGAAGATCCTCCA	774
DRD2u8	DRD2	G	A	L	L	cds	GE1088	GAGTGAGGGGTCCCTGG	CTTCGAGGGAGCAGGGG	GAGTGAGGGGTCCCTGGGCTGACCCAGATTCAGGTCCTCCCGGCTCTGGGACCAAGCTACACAGCTGTGGCCATGCCCATGCT[G/A]TACAATACGCGCTACAGCTCCAAGCGCGGCTCACCGTCAATGATCTCCATCGTGGTCTGCTCTCAACATCTCTGCGCACTCTCTCTGGACTCAATAACGCAAGTACATCTGCTTGTGTGCTGAGGCTCAGCTGGCTGGCTCCCTGGGCTCCCTCGAG	255
DRD2u9	DRD2	A	G	-	-	noncoding	GE1315	GCACCTGGGTGTGGTGTGTT	GGTCTCCAGGACTGAAGTT	GCACCTGGGTGTGGGTGTTTCCAGCGTGCCTCCCGGCTCTGGGACCAAGCTGACCATGCCCTCTCCCGAGGCGTGTTCATCATCTGCTGGCTGCCCTTCTCATCACACATCTCTGAACATACACTGTGACTGCAACATCCCGCTGTCTGTACAGCGCTTCACTGCTGGCTGTATGTCAACAGCGCCGTGAACCCCATCATCTACACCACTTCAACATTTAGTTCGCAAGGCTTCTTGAAGATCCTCCA	774
DRD3u1	DRD3	A	G	S	G	cds	GE1135	CAAGCCCCAAGGAGTCTGATTT	CGTCAACATGCACTGA	CAAGCCCCAAGGAGTCTGATTTTATTAATTTGTTTCTGCTCTCTCACAGGAAGCCCCCTTGGCACTCAAGCACTCTCTCTGGGCTATGGCATCTCTGAGTCAGCTGAGT[A/G]GCCACCTGAACACACTGTGGGAGAGAACTCCACAGTGTCCAGCGCGCCCGCCACATGCTTACTATGCCCTCTCTCTACTGCGCTCATCTCTGGGCTCTGCTTCTGGCAATGGCTGCTGTGCTGCTGTGCTGAAGGAGCGGCCCTGCACTACCACTACTTAGTAGTGAAGCTGGCTGTGGGAGACTTGTGGTGGTGGCTCACCTTGGTGTGATGCCCTGGGCTGGTGTATACCTGGAGGTGAGTAGACTTCAGGTGATGTTGACG	387

FIG. 5CC

[illegible]

FIG. 5DD

FIG. 5EE

FIG. 5FF

[illegible]

FIG. 5GG

FIG. 5HH

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
DRD5u4	DRD5	C	T	F	F	cds	GE1171	CCAGGCAGCAA CGGCAC	CTGGGCGATGC GGTAGAT	CCAGGCAGCAAACGACACGCGTACCCGGGGGCGAGTTCGCTCTATACCAAGCAGCTGGCGCAGGGGAA CGCGTGGGGGCTCGGGGCGGCGACCGCCACTGGGCGCCCTCAAGTGGTCAACGCGCTGCTGC TGACCCCTACTCATCTGGACCTGCTGGGCAACGCTGCTGGTGTGCGCAGCCACTGTCGGGAGC CGCCACCTGCGCGCAACATGACCAACGCTCTCATCGTCTCTGGCGCGTGTCAAGACCTTCTCGT GGCGCTGCTGGTCAATGCCCTGGGAGGCGAGTCCGCGAGGTGGCGGCTTACTGGCCCTTGGAGCGT TCTGCGACGTCTGGTGGCTTCTGACATCATGTCTCCACTGCTCCATCCTCACTGAACTGTG CGTCAATCAGCGTGGACCGCTACTGGGCGCCATCTCCAGGCCCTCCGCTACAAGCGCAAGATGACTC AGCGCATGGCCCTGGTCAATGGTCTGGGCGCCCTGGCATGGACCTTGTCCATCCTCATCTCTCATCCG GTCCAGCTCAACTGGCACAGGACCGGCGCCCTTGGGCGGGGCTGGACCTGCCAACAACCT GGCCAACTGGACGCCCTGGGAGGAGGACTTGGGAGCGCGAGTGAALGAGAGAACTGTGACT CCAGCCTGAATCGAACCTACGCCATCTCTCTCGCTCATCAGCTTCTACATCCCCGTTGCCATC ATGATCGTGACCTACACGCGCATCTACCGCATCGCCAG
DRD5u5	DRD5	C	A	A	A	cds	GE1171	CCAGGCAGCAA CGGCAC	CTGGGCGATGC GGTAGAT	CCAGGCAGCAAACGACACGCGTACCCGGGGGCGAGTTCGCTCTATACCAAGCAGCTGGCGCAGGGGAA CGCGTGGGGGCTCGGGGCGGCGACCGCCACTGGGCGCCCTCAAGTGGTCAACGCGCTGCTGC TGACCCCTACTCATCTGGACCTGCTGGGCAACGCTGCTGGTGTGCGCAGCCACTGTCGGGAGC CGCCACCTGCGCGCAACATGACCAACGCTCTCATCGTCTCTGGCGCGTGTCAAGACCTTCTCGT GGCGCTGCTGGTCAATGCCCTGGGAGGCGAGTCCGCGAGGTGGCGGCTTACTGGCCCTTGGAGCGT TCTGCGACGTCTGGTGGCTTCTGACATCATGTCTCCACTGCTCCATCCTCACTGAACTGTG CGTCAATCAGCGTGGACCGCTACTGGGCGCCATCTCCAGGCCCTCCGCTACAAGCGCAAGATGACTC AGCGCATGGCCCTGGTCAATGGTCTGGGCGCCCTGGCATGGACCTTGTCCATCCTCATCTCTCATCCG GTCCAGCTCAACTGGCACAGGACCGGCGCCCTTGGGCGGGGCTGGACCTGCCAACAACCT GGCCAACTGGACGCCCTGGGAGGAGGACTTGGGAGCGCGAGTGAALGAGAGAACTGTGACT CCAGCCTGAATCGAACCTACGCCATCTCTCTCGCTCATCAGCTTCTACATCCCCGTTGCCATC ATGATCGTGACCTACACGCGCATCTACCGCATCGCCAG
DRD5u6	DRD5	C	G	V	V	cds	GE1171	CCAGGCAGCAA CGGCAC	CTGGGCGATGC GGTAGAT	CCAGGCAGCAAACGACACGCGTACCCGGGGGCGAGTTCGCTCTATACCAAGCAGCTGGCGCAGGGGAA CGCGTGGGGGCTCGGGGCGGCGACCGCCACTGGGCGCCCTCAAGTGGTCAACGCGCTGCTGC TGACCCCTACTCATCTGGACCTGCTGGGCAACGCTGCTGGTGTGCGCAGCCACTGTCGGGAGC CGCCACCTGCGCGCAACATGACCAACGCTCTCATCGTCTCTGGCGCGTGTCAAGACCTTCTCGT GGCGCTGCTGGTCAATGCCCTGGGAGGCGAGTCCGCGAGGTGGCGGCTTACTGGCCCTTGGAGCGT TCTGCGACGTCTGGTGGCTTCTGACATCATGTCTCCACTGCTCCATCCTCACTGAACTGTGCGT C/G)ATCAGCGTGGACCGCTACTGGGCGCCATCTCCAGGCCCTCCGCTACAAGCGCAAGATGACTC AGCGCATGGCCCTGGTCAATGGTGGCGCTGGCATGGACCTTGTCCATCCTCATCTCTCATCCG GTCCAGCTCAACTGGCACAGGACCGGCGCCCTTGGGCGGGGCTGGACCTGCCAACAACCT GGCCAACTGGACGCCCTGGGAGGAGGACTTGGGAGCGCGAGTGAALGAGAGAACTGTGACT CCAGCCTGAATCGAACCTACGCCATCTCTCTCGCTCATCAGCTTCTACATCCCCGTTGCCATC ATGATCGTGACCTACACGCGCATCTACCGCATCGCCAG

FIG. 5II

[illegible]

FIG. 5JJ

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
F10d8	F10	C	T	F	F	cds	GE942	GGTTTTCAGTC AGGCAACAC	CAGACAGTGAC GGTGCCC	GGTTTTCAGTCAGGCAACACNagtcacacctggccagccacacactgagcctgtcagctctgtcacag gacctgtcatcaatgaggaacagaggttctgtgtggaacattctgagcgaggttctacat cctaagcgagccactgtctctaccaagcaagatt[c/t]gaaggtgaggttaggttaagt accaacagccccagggccgtgggggggggacacgctcactgctcctc
F10u1	F10	C	T	T	T	cds	GE942	GGTTTTCAGTC AGGCAACAC	CAGACAGTGAC GGTGCCC	GGTTTTCAGTCAGGCAACACNagtcacacctggccagccacactgagcctgtcagctctgtcacag gacctgtcatcaatgaggaacagaggttctgtgtggaac[c/t]attctgagcgaggttct acatcttaacggcagccactgtctctaccaagctcgaagattcggaaggtgaggttaggttaagt accaacagccccagggccgtgggggggggacacgctcactgctcctc
F10u2	F10	A	T	E	V	cds	GE907	GTCCCTCGCT GAGCTG	GTGGATCTCA CTTTAATGG	GTCCCTCGCTGAGCTGagcacagtcacacctgctcctgcccaggggacggaaacagcagcagga ggggggggtgagcggtgcacg[ta/t]ggggaggtgtcatcaagcaaacccggttcacaaag gagacctatgacttcgacatcgctgtctccggtcaagacccccacacctccgcatgaact ggcgctgctcctcccgagctgactggcgagtcacacgctcagcgtgtagcgcagacagcggga ttgtgagcgttcggggccacccacagaaagggccgagtcacacggtccacaggtcgaagtctggag gtgacctacgtggacgaacagctgcaagctgtccagcagctcactcaccagaaacatgtt ctgtgcccgtacgacacagcagcaggaagtgcctgccaggggagacagcggggccgcacgtca cccgctcaaggacacactactctgtgacaggaactcgtcagctggggagagggctgtgcccgtaa gggaagtacgggatctacacaaagtcacggcttccctcaagtggatcgacaggtccatgaaac caggggcttgccccagggcagacagcctgccccggaggtcataacgtcctcctCCATTAAAGTGAG ATCCAC
F10u3	F10	C	T	A	A	cds	GE907	GTCCCTCGCT GAGCTG	GTGGATCTCA CTTTAATGG	GTCCCTCGCTGAGCTGagcacagtcacacctgctcctgcccaggggacggaaacagcagcagga ggggggggtgagcggtgcacgaggtggaggtggtcatcaagcaaacccggttcacaaagga cctatgacttcgacatcgctgtctccggtcaagacccccacacctccgcatgaactggcg cctgctcctcccgagctgactggcgagtcacacgctgtagcgcagacagcgggaattgt gagggcttcggggccacccacagaaagggccgagtcacacggtccacaggtcgaagtctggaggtgc cctacgtggacgcacacagctgcaagctgtccagcagctcactcaccagaaacatgtcctgt gc[c/t]ggctacgacacagcagcaggaagtgcctgccaggggagacagcggggccgcacgtca cccgctcaaggacacactactctgtgacaggaactcgtcagctggggagagggctgtgcccgtaa gggaagtacgggatctacacaaagtcacggcttccctcaagtggatcgacaggtccatgaaac caggggcttgccccagggcagacagcctgccccggaggtcataacgtcctcctCCATTAAAGTGAG ATCCAC
F10u4	F10	G	C	Q	H	cds	GE250	CTGCTTCCCA GTGTTCTAT	CCCAGCCCTTA CCGCTTG	CTGCTTCCCACTTTCAGTTCacgagggagga[c/g]gccaaacacatcctggcgaggttcacgag ggccaatccttcttgaaagatgaagaaagacactcgaaagagatgcatggaagagacct gctcatcgaagggggccggaggtcttgaggacagcgaCAGACCGTAAAGCGCTGGG
F10u5	F10	C	T	N	N	cds	GE913	GAGCTTGGCTT CTGGCTA	CGTGCTCTCTA CCTGTG	GAGCTTGGCTTCTGGCTAataacgcactgcacatgagcttcccgtgacccgtgaggttgcccc ttcaaggcaagtgtacctgtgcctgtgcctgtgccccttgcacacccaatggcgcttctgtggc tgacaggcaagtgtgtgactggcacccttggccagccagcctccttctccagctgtcccc cagagccaaacgtgctccttggcagtcacacggaagctctgacgctggacaa[c/t]gggg actgtgaccagtctgcccacaggaacagacactgtgggtgtgctcctcgccgcgggtacacc ctggctgacaacggcagggcctgcatccCACAGGTAGGAGGACCG
F10u6	F10	C	A	L	I	cds	GE242	AGCCAGACAAC AGCCATCC	GGGAGGGGCAC TTACGACT	AGCCAGACAACAGCCATCCcagctggggcgtggacttctgccagcagcctgtcccagtgaggaac agggacacagtaactcggccacacactggggcgccactgcac[c/a]tcgtcctgctcagtgct ccctggctggcctcctgctgctgggggaagTCGTAAAGTGGCCCTCCG

FIG. 5KK

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
F11u3	F11	A	G	T	T	cds	GE287	TGACTTTACTTTCTCTAGGTC	ACCTCCCTCA GCATGTATC	TGACTTTACTTTCTCTAGGTCGTAaaaaatgtttttatgtgtttgatatgatatatttttacttccctttgttttgaagaaatcttctctctaaacatctgagagtggaattgcccagtaacfa/glcgcatataaagagcaaaagctcttctgttttcagttctacaaagctgcagggcacagatcccaggtaaaactgagagttctgcatctctggtgagagtgaccagccccggaggaggtctGATACATGCTGAGGAGGGT	269
F11u4	F11	A	T	II	L	cds	GE916	TGGAAGGAAGATGTAGGAAGC	GCAATAAGAC AATCTAATGG TTAAGTA	TGGAAGGAAGATGTAGGAAGCgtctcatcacaaatgtctgttgcagagtgtaaccacaaatca aagccaggatcttgagggaacacgtctgtctgtgtgagtgccgtggcaggtgaccctgca cacaacctcacccctcagagacacacgtgtgtgaggtgccatcatctggaaacacagtggaatataa cagccgctc/a/t)ctgtttctctatggttcagtaaccacggctgtttttatagttcatctcttca cacattataaaaaatattactagcatgttaggaataaaTACTTTAACCAATTAGATTGTCTTTA TTTGC	330
F11u5	F11	A	T	I	F	cds	GE339	GCCACACACTT CACAATGTC	CAGCCGCTAAG TCTAGTAGTGT TAAA	GCCACACACTTCCACANTGTCtggaattatttttttagtaaggaatcttctccctctgttctt gctccttagggtagagtcacctaagatttttgcgtgtctacagtgaggtttttaaatacattctgaaa taagaagaggaacacattcttcttgggggttcaagaataaataatcccatgatcagataaaatggca gaaagcgggtatgat(a/t)ttgcttctgttgaaactggaaacacacagtggaattacacaggtacgg agaattttatccggaaagtgtctccaatggtggaactggataaaatgtTTAACACCTACTAGACTT ACGGCTG	333
F11u6	F11	A	T	E	V	cds	GE324	CCTTTATGAGATTTACACCTAACTAGATG	TTTAAAATCT GTCTCCTCGAT GT	CCTTTATGAGATTTACACCTAACTAGATGtatgcccagtaaaaatcccaacataaacgcacccatgt actacatcacag(a/t)atgtgtgactcagttgttgaggagacacctgtttgaaaggagggggacat tactacggctcttcacacaaagccaaagtaactgcccaggtagctgcacttaccacccaagatgtt tactcttcactctcacggcggaatcaccaatctgagatcccacccagatgtaaatgttcttctgttt ctACATCGAGGAGACAGATTTTAAA	286
F11u7	F11	C	T	D	D	cds	GE283	GTATTGTGTAT GGTATTCTTAC AAACG	AAAAACCCAA CGCATTAAG	GTATTGTGTATGTATTCTTACAAACGaaacaaatatttttctcagacaaatatacaaaatact ctccagaagaacaaagataaccccttagtaccacaaagagtgccagagagatacacagagacataa aataacccataagatgatctgtgcggctacaggaagagggaggga(c/t)gcttgcaaggta acagagtgctcttagccaatggaatataatgcaaatggaaatgCTTAATGCGTGGGTTTT	257
F11u8	F11	G	A	E	E	cds	GE352	TGAGCAAGATG TUCTGAAGAT	AGCA/TGCTGGC ACAGTGAA	TGAGCAAGATGCTGTAAGATgggaagcgtctgagttgatctgtgcacctttcttctgtctccct cgtcttagggagatctgggagggcctctgtcctgcaaaacacaaatgaggtctggcatctggttaggc atcacagctggggcgaaaggtctgtctcaaaagggcgccaggtgtttacacaaacgtggtcga lg/a)ltacgtgagactggattcttgagaaactcaagcagtgtaaatgggttcccagggggccattg ggtccctgaaggacccaggatttctgtggagaggtgttgaggtTCCAGCATGCT	322
F11u9	F11	T	C	L	P	cds	GE324	CCTTTATGAGA TTACCACCTAA CTAGA	TTTAAAATCT GTCTCCTCGAT GT	CCTTTATGAGATTTACACCTAACTAGATGtatgcccagtaaaaatcccaacataaacgcacccatgt actacatcacagaaatgtgtgactcagttgttgaggagacacacctgtttgaaaggaggagacattact acggctcttcacacaaagccaaagtaactgcccaggtagctgcacttaccacccaagatgtttac l/c)cttcactctcacggcggaatcaccaatctgaggtatcccacccagatggtaaatgtcttattgttt ctACATCGAGGAGACAGATTTTAAA	286
F13A1d 24	F13A1	C	T	-	-	noncoding	GE284	TGCCATGAATC TTGCAGATC	CAACTTTTAGC TTACTCTTTCA TGTG	TGCCATGAATCTTGCAGTATCtgggacagaatggagatgacaaactcaca(c/t)tgcccttccct ctgtgcaatgcaggcatgtatcgggtggtggcccgccctcggttcaggccatcaagcacggccatgt ctgcttccaatllyatgcacctttgttttgcagaggtgaagcaggaagctggaggaaagtgctg tttctctcccaacctcttaacACATGAAGAGTAAAGCTTAAAGTTG	242
F13A1d 25	F13A1	T	C	-	-	noncoding	GE284	TGCCATGAAT TTGCAGTATC	CAACTTTTAGC TTACTCTTTCA TGTG	TGCCATGAATCTTGCAGTATCtgggacagaatggagatgacaaactcaca(c/t)tgcccttccct ctgtgcaatgcaggcatgtatcgggtgtggcccgccctcggttcaggccatcaagcacggccatgt ctgcttccaatllyatgcacctttgttttgcagaggtgaagcaggaagctggaggaaagtgtg tttctctcccaacctcttaacACATGAAGAGTAAAGCTTAAAGTTG	242

FIG. 5NN

[illegible]

FIG. 500

[illegible]

FIG. 5PP

[illegible]

Poly Id		Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
F13A1u 8		F13A1	T	C	D	D	cds	GE344	GCCCCTACAAAG AAGGGTTTT	AGAACAGAAAC ATCAGATTGAG TCTA	GCCCCTACAAAGGGTTTTTcactctgtgttattataatgctgggtgatgtgtttagctgtggt ctgtctcttctgttagttttacgatgccttggaatcacacgcaagaattgttaccatattttctc tgcccatga [t/c]aatgatgccaatgttgcgaatggacatcttctctggaagaagatgggaacgtg aatcccaactcaccacgaagattcagttggtgagtttgaatgcaaatggcatggctgagtcac attatagtaattgttacacattaccacaagatAGACTCAATCTGATGTTTCTGTTCT
F13A1u 9		F13A1	C	A	P	T	cds	GE487	ACACGGTGCAT CCATTA	CGAGTCTCACA NAGAACCA	ACACGGTGCATCCATTAatgtgactttctctctgggtggattgtattttgacctgtcattctct ggatctccccccagggtcaagaagaagagagattggccctagaacacccctgattgacggagctaa aaagccctcaacacagaaggtgtcatgaaatcaagggtccaacttgacatggactttgaagtgg aaaaatgctgtgctgggaaaagacttcaagctctctcatcacttcggaacacacagccacacccg tacaccatcacagcttattctctcagccaacatcacttctacacgggggtc [c/a]cgaaggcag aatccaagaaggagacgttcgacgtgacgctggagcccttgcctctgaagctaaccaatgggtg TTCTTTGTGAGACTCG
F13Bd2 0		F13B	T	G	-	-	noncoding	GE264	AAATTTGAAAG GCTGAGATTGT AA	CAATATTTTAA GCAAGGAAAA CTCC	AAATTTGAAAGGCTGAGATTGTAA [t/g]taacacotgactgcaattgatgttatttcaaaaat ctctcttttccctcaagcactctgtcttatacaagaaccccttaagaacatagaaaatgaatgga gaagaggagtcataattcaatcacatcatgaaatctctataaaaataaatttttgaggaaalaag tcaaaaacttcggactTTTTTCTTCTTCTTAAATATTG
F13Bd2 1		F13B	A	T	E	V	cds	GE370	CTTGAAAGAGC TTTGCTAAAT G	GCCTAAGCAGT GGTCTTTTCT A	CTTGAAAGAGCTTTGCTAAATGaaatctgcagctgtagctaaatggctcctcttcaactg taccttttcagaagacagagaggtagcctgtggaagaaaccccttcatgaaaatgggtgcag caaatttacactcaagattattacaatggggaataaagtgacatattgcatgaaaagcggctac cttccatggatcgaaatgagataacttgaatctggtggaatggacacttctctctg [a/t]gt gtgtgggtatgtatgctacatttaccatcagtagtaacttattggtgtaaaatttccactctg ttactttaaactctgaatattctTAGGAAAGACCCTGCTTAGCC
F13Bd2 2		F13B	A	G	Q	Q	cds	GE373	CCAAATGAAA TCGCCAATAAT A	TTCTCTGCATTG TAGACATAATG A	CCAAATGAAATCGCCCAATAATAacattataacttttggcttaatttacaatttagtaaaagaca agcttagtttcatcattcaagtttaaatattttttcccatagaaaatgcactaagcctgacct gagtaatgggttacatctctgtatgaagtatttgtataaaattca [a/g]gagaacatgcattat ggttgcgcttcagggtacaaaaccactggagggaaggaaggaaggttcaatgtctctctga tggaatgggtcttctcaaccacccctgtaggaaagaaacatgggtataagaatcatttccctaaanctgaa gataagtgcttatacgaagcttatttgaatatatttgaagcttatttgatatttatttattatag aatattatttTCATTATGCTTACAAATGACAGAA
F13Bd2 3		F13B	T	C	-	-	noncoding	GE373	CCAAATGAAA TCGCCAATAAT A	TTCTCTGCATTG TAGACATAATG A	CCAAATGAAATCGCCCAATAATAacattataacttttggcttaatttacaatttagtaaaagaca agcttagtttcatcattcaagtttaaatattttttcccatagaaaatgcactaagcctgacct acctgagtaatgggttacatctctgtatgaagtatttgtataaaattcaagaagaacatgcattat ggttgcgcttcagggtacaaaaccactggagggaaggaaggaaggttcaatgtctctctga tggaatgggtcttctcaaccacccctgtaggaaagaaacatgggtataagaatcatttccctaaanctgaa gataagtgcttatacgaagcttatttgaatatatttgaagcttatttgatatttatttattatag aatattatttTCATTATGCTTACAAATGACAGAA
F13Bd2 4		F13B	T	A	-	-	noncoding	GE373	CCAAATGAAA TCGCCAATAAT A	TTCTCTGCATTG TAGACATAATG A	CCAAATGAAATCGCCCAATAATAacattataacttttggcttaatttacaatttagtaaaagaca agcttagtttcatcattcaagtttaaatattttttcccatagaaaatgcactaagcctgacct gagtaatgggttacatctctgtatgaagtatttgtataaaattcaagaagaacatgcattatggtt gcgcttcagggtacaaaaccactggagggaaggaaggaaggttcaatgtctctctgga tggtcttctcaaccacccctgtaggaaagaaacatgggtataagaatcatttccctaaanctgaa agtgcttatacgaagcttatttgaatatatttgaagcttatttgatatttatttattatag aatattatttTCATTATGCTTACAAATGACAGAA

FIG. 5RR

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
F13Bd2 5	F13B	C	T	C	C	cds	GE385	CACCTTTTCATGCATTTTTCGC	TGACCACAGGA ATTTTGTCCAGA	CACCTTTTCATGCATTTTTCGCctttatttttaattatgtctgtagcagctcttttgataaattaaagc tctcaagtagaataaaattcatttttagctcacagattaaattagataaagcctaaatgatatttaa caaatataaaatgaattcataagcttttttgcaaatataattctattcttagttagtcatgt aaatagtcatttttggtagcagaaaaataaacatttggttgaattttattctctcttttat ctgcagaattaaagtgcctctcttaagattaaatgaaattggatttttcatctctgtaagcaa acctatgaagaaggagatgctgtcagttttctgtgcatgaaattattatctaaaggatctga tttaattcaatg[c/t]ataactttgggtgggtaccagaaattctctgtatgcgaaggtaaaattt ttaaatttaatactcgaataatgccatgtctttatcagctttaatctctatttagctCTGACAAAA TTCTGTGGTCA
F13Bd2 6	F13B	T	C	-	-	noncoding	GE396	TGTTGGATGTT TAAAGTCATTT GC	CATTTTATTG GACCCCTATT T	TGTTGGATGTTTAAAGTCATTTGCagatcaattatgataaaaggactccttgagttgtcacaaaa gtacctaaaaatttaagtaagaaaaataagtagtactagttgaaagtgtctctaaaaacttttagttt gtattgtcacctgatcacaaatttatgttttttagatttggttt[c/c]ccattatcagcttgaca tttactcatttcagagaaaacctgtggtttctctcatgtggaaaaatggaaatattgcccaatttt actatacttttaaaagcttttactttccaatgagcatagacaaaaatgtcatttttctgtctg gctgttataccactgaaagtggaaagacaagaaacacacgtagacaagaaaggtggtc tccagagccaaggtgcttcagtaagtcagctggatgtgtcactcaatgtttcaatactcaagaa attgtatatataAAATAGGGGTCCCAATAAAATG
F13Bd2 7	F13B	T	A	-	-	noncoding	GE396	TGTTGGATGTT TAAAGTCATTT GC	CATTTTATTG GACCCCTATT T	TGTTGGATGTTTAAAGTCATTTGCagatcaattatgataaaaggactccttgagttgtcacaaaa gtacctaaaaatttaagtaagaaaaataagtagtactagttgaaagtgtctctaaaaacttttagttt gtattgtcacctgatcacaaatttatgttttttagatttggtttccattatcagcttgacattta ctcatttcagagaaaacctgtggtttctctcatgtggaaaaatggaaatattgcccaattacta tacttttaaaagcttttactttccaatgagcatagacaaaaatgtcatttttctgtctgctg gtataccactgaaagtggaaagacaagaaagcaccacgtagacaagaaaggtggtctcca gagccaaggtgcttcagtaagtcagctggatgtgtcactcaatgtttcaatactcaagaaattt gtatat[c/a]AAATAGGGGTCCCAATAAAATG
F13Bd2 8	F13B	C	G	-	-	noncoding	GE396	TGTTGGATGTT TAAAGTCATTT GC	CATTTTATTG GACCCCTATT T	TGTTGGATGTTTAAAGTCATTTGCagatcaattatgataaaaggact[c/g]cttgagttgtcac aaaagtaaccttaaaaaatttaagtaagaaaaataagtagtactagttgaaagtgtctctaaaaactttta gttgttatgtcacctgatcacaaatttatgttttttagatttggtttccattatcagcttgaca tttactcatttcagagaaaacctgtggtttctctcatgtggaaaaatggaaatattgcccaattat actatacttttaaaagcttttactttccaatgagcatagacaaaaatgtcatttttctgtctg gctggttataccactgaaagtggaaagacaagaaagcaccacgtagacaagaaaggtggtc tccagagccaaggtgcttcagtaagtcagctggatgtgtcactcaatgtttcaatactcaagaa attgtatatataAAATAGGGGTCCCAATAAAATG
F13Bd2 9	F13B	G	A	T	T	cds	GE481	CAATTTTGCCA AGAGTTGATT	TTCTCTATGAG AAAAAGCTTTC AG	CAATTTTGCCAAGAGTTGATTTtaccatgtattcttaagatttgaataaccatatttttgagattt tttataatctaggttatataaaagtttataaaaaataaatttttataatttttagaaac[g/a]t gtttggctcctgaattataataatggaattattcccaacacagaaaaacattcaagtgaaaggac aaagtaacaatcgaattgtctactggtcactacacagctggaggaagaaagacagagggtaga atgtctcaatagcggatggctctcacacaaaatgtaccagtaggtttctcatttggaaataaaat ctactctcactCTGAAAGCTTTTCTCATAGAGAA
F13Bd3 0	F13B	A	G	-	-	noncoding	GE481	CAATTTTGCCA AGAGTTGATT	TTCTCTATGAG AAAAAGCTTTC AG	CAATTTTGCCAAGAGTTGATTTtaccatgtattcttaagatttgaataaccatatttttgag attttttataatctaggttatataaaagtttataaaaaataaatttttataatttttagaaacgt gtttggctcctgaattataataatggaattattcccaacacagaaaaacattcaagtgaaaggac aaagtaacaatcgaattgtctactggtcactacacagctggaggaagaaagacagagggtaga atgtctcaatagcggatggctctcacacaaaatgtaccagtaggtttctcatttggaaataaaat ctactctcactCTGAAAGCTTTTCTCATAGAGAA

FIG. 5SS

FIG. 5TT

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
F13Bu1 6	F13B	T	C	P	P	cds	GE370	CTTGAAAGAGC TTTGCTAAAT G	GCCTAAGCAGT GGTCTTTTCCT A	CTTGAAAGAGCCTTGGTAAATGaaatctgcatgtgtagtaaatggctgaatggctctcttccaactg taccttttcagaagagacaggagagaggtgagcctgtgaggaacacccttccatgaaatggtgcag caaatctacactctaaagatttattacaatggggataaagtgcacatatgcatgtaaaagcggctac ctctccatggatcgaaatgagataaacttgtaactggtggaatggacacttcc[t/c]cctgagt gtgttggtatgtatgtacatttaccatcagtagtcaacttatggtgtaaaatttccattctg ttacttcaatctgaatatctcTAGGAANAGCACCCTGCTTAGGC	369
F13Bu1 7	F13B	T	A	N	K	cds	GE400	TTTGCTGCTCAA CTCTGCTTAG	AAATGAGAGAG AAAACACTTGT GAAA	TTTGCTGCTCAACTCTGCTTAGGcaaatcttcaaaatatttgagctgaaaaatttccatatttg taatgaatttataaaacaattgtcctcaattataataatacaaaatcgagacactatagtagag ttactatagtatatgttatgctcctcatatlaatttcaaaacaaccttcttcttagaaaaa[t/a laatggaattgtgaagcatcctcctgtgtgaatgaatgggctgttgcagacgggatattggcaa gctatgcaacaggatcctcagtggaatatagatgcaatgcaatattactactgagggatcaaaa atatctcgttgcgaacaaggaaaatggcatccccacctgttgcctggglaagaaagagaaacac atggaatgtcagcttcttacttcttctgatttcttctacaggttttatcatcatgaaaaatgatg atttctgtaaaaatcttcttCACAAAGTGTTCCTCTCTCATTT	498
F13Bu1 8	F13B	G	C	L	L	cds	GE264	AAATTTGAAAG GCTGAGATGT AA	CAATATTTAA GCAAGGAAAA CTCC	AAATTTGAAAGGCTGAGATTTGTAattacacctgactgcaattgatgcttatttcaaaaatctct cttttccccctcaagcactct[g/c]tcttatacaagaaccttgaagacatagaaatgaatggca gaagaggagatcatattcaatcacatgaaatctctataaaatataatttctgaggaataaag tcaaaaactctGGAGTTTTCTCTGCTTAAATAATTG	232
F13Bu1 9	F13B	T	A	N	K	cds	GE400	TTTGCTGCTCAA CTCTGCTTAG	AAATGAGAGAG AAAACACTTGT GAAA	TTTGCTGCTCAACTCTGCTTAGGcaaatcttcaaaatatttgagctgaaaaatttctcatatttg taatgaatttataaaacaattgtcctcaattataataatacaaaatcgagacactatagtagag ttactatagtatatgttatgactcatatlaatttcaaaacaaccttcttcttagaaaaataa[t t/a]gagaattctgaagcatcctcctgtgtgaatgaatgggctgttgcagacgggatattggcaa gctatgcaacaggatcctcagtggaatatagatgcaatgcaatattactactgagggatcaaaa atatctcgttgcgaacaaggaaaatggcatccccacctgttgcctggglaagaaagagaaacac atggaatgtcagcttcttacttcttctgatttcttctacaggttttatcatcatgaaaaatgatg atttctgtaaaaatcttcttCACAAAGTGTTCCTCTCTCATTT	498
F13Bu2	F13B	A	G	M	V	cds	GE396	TGTTGGATGTT TAAAGTCATT GC	CATTTTATTTG GACCCCTATT T	TGTTGGATGTTTAAAGTCATTGcagatcaattatgataaaaggactccttgagttgtcaca gtaccttaaaaatttaagtgaagaaataaagtactagtgaagtgctcttaaaaacttttagttt gtattgtcacctgattacaaatttatgttttttagatttgttttccatattcagcttgacattta ctcatctcagagaaacccctgtgttctcctcatgtggaataatggagaattgcccaatattacta tacttttaaaagcttttacttcca[a/g]tgagcatagacaataatgtcattttctctgcttg gcgggttataccactgaaagtgaagacaagaagagacacccagctgtacaacagaagcctggct tccagagccaaaggctgtcagtaagtcagctggatattgtcactcaatgtttcactactcaagaa atttctgatatataAAATAGGGTCCCAATAAATAATG	489
F13Bu3	F13B	A	G	H	R	cds	GE373	CCAAATGAAA TCGCCAATAAT A	TTCTGCTATTG TAGACATAATG A	CCAAATGAAATCGCCAATAATAacattatatttctggcttcaatttcaaaatttagtanaagaca agcttagtttccatcattaaagttaataatatttctccatagaaaatgcactaagcctgacct gagtaaatggtttacatctctgattgaagtatttctataaaaatcgaagagaacatgc[a/g]ttat gggtgcgcttcagggtacaaaacccactggagggaggtgaagaagtgggttcaatgtctctctga tggatggctctctcacaacccctgtaggaagaaacatggtataagaatcatttctcaaanctgaa gataaagtgtctatcaagcttatttgaatatattgaagcttattgatatctattttaaag aatttatttctCATTTATGCTCAATGCAGGAA	423

FIG. 5UU

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
F13Bu4	F13B	A	T	N	Y	cds	GE358	GATGCTTGACA CGATGAATATT TTA	GTACAGTTGAA AGAGAACTGAC CA	GATGCTTGACACGATGAATATTTTAAatttaattcttatttttctgtaattalatgactaaagg lgctgatttaatttttccataaggaagaaacagatgctccctccaccctctgcccataaact ccaaatttcaaaacacattcaacaacttattctgcatgagagaattagttcatalagaaatgtgaactt [a/c]attttagatccatgggtcagcagaatacgttctgaagatggaatcgacagaacctc caaaatgcattggtagtaaacaccttgaagaagcttctgctaaatgaaatcgcacgtgtgtagta aaTGGTCACTGCTCTTCAACTGTAC
F13Bu5	F13B	C	T	P	S	cds	GE370	CTTGAAGAAGC TTTGCTAAAT G	GCCTAAGCAGT GGTCTTTTCCT A	CTTGAAGAAGCTTTGCTAAATGaaatctgcatgctgtagtaaatggcagttctcttccaactg taccttttcagaagagcaggaagaggtgctgtaggaaccacaccttcatgaaatgggtgcag caaatttacactctaaagatttattacaaatggggatgaagtgacataatgcatgtaaaagcgctac cttctccatggatcgaaatgagataaacttgaatcgtggaataattactactgaggggatcaaaa gtgttggtatgtatgctacatttaccatcagtagctaaacttattggtgtaaaallttccattctg ttactttaattctgaatatctcctAGGAAGACCACTGCTTAGGC
F13Bu6	F13B	T	A	M	K	cds	GE400	TTTGCTGTCAA CTCTTGCTTAG	AAATGAGAGAG AAACACTTGT GAAA	TTTGCTGTCAACTCTTGCTTAGcaaatttttaaataattttagctgaaaaaatttccatatttg taatgaatttataaaaaaattgctctaaattataatacaaaatcagagacacactatagtagag ttactatagtagtattgtttagctcattataattttaaacaacctcttcttagaaaaataat gagaattgtaagcattcctctctgtgtgaatgaatgggctgttgcagacggga[t/a]atttgcca gctatgcaacaggatcctcagtggaatatagatgcaatgaattactactgaggggatcaaaa atactcgttgcgaacaaagaaatggctcaccacctgttcttgggtgaagaagagagacac atggaatgctacgttctgacttttattgtgattttctacagtttttatcatgaaatgatg atttctgtaaaatcttTTTACAAAGTGTCTCTCTCATTT
F13Bu7	F13B	T	A	I	K	cds	GE400	TTTGCTGTCAA CTCTTGCTTAG	AAATGAGAGAG AAACACTTGT GAAA	TTTGCTGTCAACTCTTGCTTAGcaaatttttaaataattttagctgaaaaaatttccatatttg taatgaatttataaaaaaattgctctaaattataatacaaaatcagagacacactatagtagag ttactatagtagtattgtttagctcattataattttaaacaacctcttcttagaaaaataat gagaattgtaagcattcctctctgtgtgaatgaatgggctgttgcagacggga[t/a]atttgcca gctatgcaacaggatcctcagtggaatatagatgcaatgaattactactgaggggatcaaaa atactcgttgcgaacaaagaaatggctcaccacctgttcttgggtgaagaagagagacac atggaatgctacgttctgacttttattgtgattttctacagtttttatcatgaaatgatg atttctgtaaaatcttTTTACAAAGTGTCTCTCTCATTT
F13Bu8	F13B	G	A	-	-	noncoding	GE400	TTTGCTGTCAA CTCTTGCTTAG	AAATGAGAGAG AAACACTTGT GAAA	TTTGCTGTCAACTCTTGCTTAGcaaatttttaaataattttagctgaaaaaatttccatatttg taatgaatttataaaaaaattgctctaaattataatacaaaatcagagacacactatagtagag ttactatagtagtattgtttagctcattataattttaaacaacctcttcttagaaaaataat gagaattgtaagcattcctctctgtgtgaatgaatgggctgttgcagacggga[t/a]atttgcca tgcacaggatcctcagtggaatatagatgcaatgaattactactgaggggatcaaaaataat ctcgttgcgaacaaagaaatggctcaccacctgttcttgggtgaagaagagagacac atggaatgctacgttctgacttttattgtgattttctacagtttttatcatgaaatgatg atttctgtaaaatcttTTTACAAAGTGTCTCTCTCATTT
F13Bu9	F13B	T	A	-	-	noncoding	GE380	GGAGACTCTGT CTCTGTAGCAC TTAT	AACACAAATGT AGCAATATAG CAT	GGAGACTCTGTCTGTAGCACTTATAaagtagtggatgttccattatagcaattcattgtatact ttaaaact[t/a]atttttgcagaatctaaaggaatgtgcacatctctcctcttattaaacatg gagtcattattagttcaacagtagacacctatgaaaatggctcttcagtgaatacagatgtttt gatcaccatttcttagaagatctagggggacctattgttttagatggaatgtggaactacaccac attgtgttttaggtatgtactactaaatattgcctcttaacaaagtaaaactatatttttaatttg ctgtcatttttgagttaaacataaacactaatatAATGCTATATTGCTACATTGCTGTT

FIG. 5VV

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
F5d47	F5	G	A	-	-	noncoding	GE949	TCCCTAACCATGGAGTTTACTTTATTG/a tatatatatttataagcaaaattattcattttt GGAGTTTACTTT	GCACAACTGTA GGGGTA	TCCCTAACCATGGAGTTTACTTTATTG/a tatatatatttataagcaaaattattcattttt gatcaaggcttcagagtttctgggtaagttagcacaatggtctgtaattggtTACCCCTACAGT TGTC
F5d48	F5	A	G	-	-	noncoding	GE316	GCAAAGGTTT AACATCTTCT	GCACAGTCTTC AGATTGCTTT	GCAAAGGTTTAACTTCTTATctatgtgttttttgcattttc(a/g)taggcaaaacaaca taagcagtggttagaatttgatctactcaagatcaagaagataacggcaattataacacagggct gcaagctctctgctctgtaaatgtagtaagagctataaccctccactacagtgagcagggagtg gaatggaaaccatacaggtgaaatctctccatggtagacaaggtagagtggtcatctgggcaaaaga GAAAGCAATCTGAAGACTGTGC
F5d49	F5	C	G	H	Q	cds	GE387	GTGCCCCCAG GGAACTACTA	TCATATGGCTG AGTTCTGGAG	GTGCCCCCAGAGGAACTACTAcaaacattccccattcaagaccctgatcaaatgcactctacttc agacccccagtc(a/c/g)agatctctctccagagctcagtgaaatgcttgagatgacccgaagt cacaagctctcccccagagataaagtcaaatgtccccctctccagaacatgaagtctggcagac agtcattctccagaccctcagccaggtgacctctctccagaactcagccagcaaaacctctctc cagacctcagcacacagctctctccagaaactcattcagagaaaccttccccagccctcggt cagatgcccatttctccagacctcagccatacaaccttctccagacctcagccatacaacct tcttttagacctcagccagacaacctctctccagaactcagtcagacaaccttctccagccc tcggtcagatgccccttctccagacctcagccatacaaccttctcttagacttcagccagaca aacctctCTCCAGAACTCAGCCATATGA
F5d50	F5	A	G	T	T	cds	GE389	GAAAGGTAGCT ATGAATAATC CAAGA	GGGTCTTGAAT GGGGAATCT	GAAAGGTAGCTATGAATAATCAAGTactgatgaagacacagctgttaacaattggctgataca gccccagaatgcctcagctgcttgggagaaagcaccctcttgccaacacagctggaaagcag agtggccaccacaagtctcttagayttagacataaattctacaagtaagccaggtggaggaaa gagttagactgaagaaagccagtttctcatttagacacacgcaaaagaaagagagacacac acctgctcttctctccagagaccttccacctctcaagagtgagcctacaacacatttca gaaagaagacttaagcattcgttggtgcttcataaattccaatccaatgaaacatctctccacagacct caatcagac(a/g)ttgcccctctatggaatttggctggatagcctcacttctgaccataatcag aatctctcaaatgacactggtcaggaagctgtctccaggtctttatcayacagtgccccccaga ggaacactatcaaacATTCCCATTCAGACCC
F5d51	F5	A	C	E	A	cds	GE496	CTTCTCCAGA CCTTGCTGAG	AAGGAGAAAC TGCCCAAC	CTTCTCCAGACTTGGTGAGacagatcttccccaaaactttggtcagatgtcccttccccaga cctcagccaggtgactctctccagacatcagtgacacaccttctccggatctcagccaga tatcactctccagacctgagtcagatatctccctctgaaatctagtcagtcattgctctct caagbaatttaagagctcttctctccagaccttggtcagatgacctctctctctctctac tctcaatgatacttctatacaaaaggaatttaaccactggttatagtgggcctcagtaaaagatg gtacagattacattgagatcattccaaaggaag(a/c)ggtcagagcagtgagatgactatgc tgaaattgattatgtgcccctatgatgacccctacaaaactgatgttaggacaaacatacaactcct ccagagatcctgacaacattgcagcaggtgacctccgagcaacaatggaaacagaaatattat tacattgctgctgaagaaatattcctgggattattcagaatttgtacaaagGTTTGGCCAGTTTTC TCCTT

FIG. 5WW

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
F5d52	F5	C	A	-	-	noncoding	GE910	GCAGGGAATAT TGGTGTG	TCTACTGATAC AATGGCAGAGT T	GCAGGGAATATTTGGTGTGtaataatca(c/a)ggaaccaccacatgacaggatgttccacacc cctgggtatggaatggaagatagaaacaaacaaacacagcttctcggttaagaatcttct ggtagagattactgggaacccctccgtgccgtctgaatccccaggagcgtgtgaatgcctgg caagcaagggtcaagataactctatgcattggttctcttagggctcccaaaaaaagcaaggcc ctactaccacaacacatggaggatctgggaagcaggagtatttaactctgttctgcaaattaaa atcctagggtccaanagtcaggatgtctgtggacttgataacttgaactataaacttctctcaaa aanaagtcatttatactatctctgtgtgttctgtgtctatctctgatttaaaccaataataagcaaat taatttctctgcaaggatanaatggaatatttttagtcttctgtggccatacatctctgcca taactactcaactcgaactgccaattgctatcagtaaa	552
F5d53	F5	C	T	-	-	noncoding	GE915	TGATTATCAGA AGAGCAAGGAA A	ACTGTGACCCA GTGTGATTTA	TGATTATCAGAGCAAGGAAAtcctgagaaagggaatacaaatctactctgttttccagg aaaaagataattcaactcaggcttgataggtccccctctaatctgccaaaaagggaata-tacataag gacagcaacatgcctatggacatgagagaatttgtcttactatttatgacctttgatgaaaaaa gagctggactatgaaaaaaggccccgaagttcttggagactcacatctcagaaaatgaaaaaat cccatgaggttccacgggtatttctct(c/a)ggacttggatctaatctcctaatTAAATCACACTG GGTACACAGT	334
F5d54	F5	A	G	-	-	noncoding	GE915	TGATTATCAGA AGAGCAAGGAA A	ACTGTGACCCA GTGTGATTTA	TGATTATCAGAGCAAGGAAAtcctgagaaagggaatacaaatctactctgttttccagg aaaaagataattcaactcaggcttgataggtccccctctaatctgccaaaaagggaata-tacataag gacagcaacatgcctatggacatgagagaatttgtcttactatttatgacctttgatgaaaaaa gagctggactatgaaaaaaggccccgaagttcttggagactcacatctcagaaaatgaaaaaat cccatgaggttccacgggtatttctct(c/a)ggacttggatctaatctcctaatTAAATCACACTG GGTACACAGT	334
F5d55	F5	A	G	G	G	cds	GE923	GATCATTTCTT TTCCTAGGTT	TTCAGATTACG AGGTTAGGGGA	GATCATTTCTTTTCTTAGGTTTggttttaaaaaatttagcatccagaccgtattctctacatgccca tgg(a/g)cttctctatgaaaaatcatcagagggaagacttaagaagatgactctctcctgaatgg tttaagggaagataatgctgttcagccaaatagcagttataacctcgtatggcatgccactgagcg atcagggccagaaagtcctgctcctgcctgcggccttgggctactactcagctgtgaaccag caggtaacttcatgaaagtcttctctcatTCCCTAACCTCGTAATCTGAA	311
F5d56	F5	C	G	T	S	cds	GE923	GATCATTTCTT TTCCTAGGTT	TTCAGATTACG AGGTTAGGGGA	GATCATTTCTTTTCTTAGGTTTggttttaaaaaatttagcatccagaccgtattctctacatgccca tggacttctctatgaaaaatcatcagagggaaga(c/g)ttatgaagatgactctcctgaatgg tttaagggaagataatgctgttcagccaaatagcagttataacctcgtatggcatgccactgagcg atcagggccagaaagtcctgctcctgcctgcggccttgggctactactcagctgtgaaccag caggtaacttcatgaaagtcttctctcatTCCCTAACCTCGTAATCTGAA	311
F5d57	F5	G	A	-	-	noncoding	GE924	TTGCTTGCCTT AAACCTCTTG	GAAACAGGAC CGAAAAATTAC TA	TTGCTTGCCTTAACCTTTTgntatccaggaagaantaaatgagn(g/a)tnntttatctgcagtgc tactgaaaaacaagctcttctcttcatagggacccttaactgagggtgggacacagaagacgttga caagcaaatcgtgctactatttgcgtgtgttgcagtgaagcaagagctggagccagtcacatccc taatgtacacagtcgaatggatgtgtaattgggacaatccaggtaaacacaggggctgtgtaccat taacaacagtaaaatcatTAGTAATTTTTCGCTCTGTTTC	303
F5u1	F5	C	T	N	N	cds	GE173	AAATTTGATTT AACCTTTGTAGA TCCGTG	GGATTTCAGTAG AAGTGAAGAT TCAA	AAATTTGATTTAACCTTTGTAGATCGTGTtcaaaaaataggccagcccccctatagcatttacc ccatggagtgacctctctcctctatgaagatgaagtc(a/c/t)cttcttctcacctcaggtTTG AATCTTTCACCTTCTACTGAATCC	153

FIG. 5XX

[illegible]

FIG. 5YY

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
F5u15	F5	T	G	S	A	cds	GE395	GACCTCAGCCCA GACAAACCT	TCTGATCAAGG TCTGGAGGA	GACCTCAGCCCAACCAACCTCTCTCCAGAACTCAGTCAGACAAACCTTCTCCAGCCCTCGGTCA gatgccccttctccagacctcagccatacaaaccttctctagactcagccagacaaacctct ctccagactcagccatacagctctctctccagactcagtcagacaaaccttctccagccctc ggtcagatgccattctccagacctcagccatacaaaccttctctagactcagccagacaaa cctctccagactcagtcacaaaaccttctccagccctcagtcagacaaaccttctccag acccagccatacaaaccttctctagactcagccagacaaaccttctccagaaactcagtcag acaaaccttctccagacctcagtcagactcagccctctctcagactcagtcagaaactcccttac cccagacctcagacagatgacacttctccagacctcagtcagactcagtcagacagacttctg cccccaaac tctggtcagatgccccttctccagacctcagccagctgactctctctccagacatcagtcagacac cacccttctccagactcagccagatataccctTCTCCAGACCTTGATCAGA	638
F5u16	F5	C	A	S	S	cds	GE496	CTTCTCCAGA CCTTGGTGAG	AAGGAGAAAC TGGCCAAAC	CTTCTCCAGACCTTGGTGAGACAGATCTTCTCCCAACCTTGGTcagatgccccttctccag cctcagccaggtgactctctctccagacatcagtcagacacaccttctccggatctcagccaga tatcacctctccagaccttggatcagatatacccttctgactctcagtcagtcagtcagtcctt caagaaattaaatgagctcttctcttccagaccttggtcagatgccatctctctcctctac tctcaatgataacttctatacaaggaatttaaccactggttatagtgggcctcagtaaaagatg gtacagattacattgagatcattccaaaggaagaggtccagagcagtgaaagatgactatgctgaa attgattatgcccctatgagacccctcaaaactgatttaggacaaacatcaactc c/a t ccagagatcctgacacattgacagatggtacccctcagcagcaaatggaaacagaaacattat tacattgctgctgaagaaatactcctgggattattcagaatttgcacaaaggtTGGCCAGTTTTC TCTT	590
F5u17	F5	A	T	N	I	cds	GE116	GCAAGGTTT AACATCTTCCT T	GCACAGTCTTC AGATTGCTTT	GCAAGGTTTAAACATCTCTCTTatctatgtgttttctgctatttcataggcaaca a/t caa taagcagctggctagaaattgactctactcagatcaagaagataacggcaattataacacagggct gcaagtctctgctcctgaaatgtagtgaagagctataccatccactacagtgagcagggagtg gaatggaaacccatcacggctgaaatcctccatggtggacaaggtagagtgccatctgggcaaa gAAAGCAATCTGAAGACTGTGC	282
F5u18	F5	G	C	D	H	cds	GE267	TGATGACCCCTG AATACAGACAT AGT	AGTTTAAATG TTGATGCTGGT ATT	TGATGACCCCTGAATACAGACATAGTTtaaatatttggtttccacaggactcttgggcccacttta tatgctgaagtcggagacatcataaaagtctcacttttaaaaaaaggca g/c ataagcccttga gcatccatcctcaaggaaattaggtcacagtaattatcagaaggtgaagataaatctaatatttctc tgaaaaagggaattgcttaagctcttAATACCAGCATCAGATTTAAACT	246
F5u19	F5	G	A	K	K	cds	GE267	TGATGACCCCTG AATACAGACAT AGT	AGTTTAAATG TTGATGCTGGT ATT	TGATGACCCCTGAATACAGACATAGTTtaaatatttggtttccacaggactcttgggcccacttta tatgctgaagtcggagacatcataaaagtctcacttttaaaaaaaggcagataa g/a cccttga gcatccatcctcaaggaaattaggtcacagtaaatatcagaaggtgaagataaatctaatatttctc tgaaaaagggaattgcttaagctcttAATACCAGCATCAGATTTAAACT	246
F5u2	F5	G	A	R	K	cds	GE47	GAATAACTTT GCAATGAAA CA	TTTCTGAAAGG TTACTTCAAGG AC	GAAATAACTTTGCAATGAAAACAACTtttgaatataatttcttccaggcaggaacaacacatga tcagagcagltcaaccagggggaaacctatactataagtggaacatcttagagttttagtaacccc acagaaaaatgatgcccggtgcttaacaagaccatactacagtgacgtggacatacga g/a ag acatcgccctctgggctaataggactactcttaattctgtgaagacagatcccttgacagggcagga atacagggtatttTGCTCTGAAGTAACCTTTTCAGAA	297

FIG. 5ZZ

[illegible]

FIG. 5AAA

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
F5u26	F5	C	T	-	-	noncoding	GE924	TTGCTTGCCCTTAACTCTTTGNTATCAGGAGAAAGTAAATGAGNGTNTTTTATCTGCAAGTCTACT	GAAACAGGACCGAAATAATTAC TA	gaaacaagctcttctcttctcatagggagccctaaactgaggggtgggacacagaagagcttggacaag caaatcgtgctactatttgcgtgtgttgatgaaagcaagagctggagccagtcacatccctaatgtacacagtcgaatggatattgtaattgggacaatgccaggttaaca/c/t gagggctgtgtaccat taacaacagtaaaatcatTAGTAATTTTTCGGTCTCTCTTTTC
F5u27	F5	A	G	E	E	cds	GE494	TCTGTCTCTCTCTCTGTGTAGGAAC	CCCTGTGACATCTGGCTGTAG	TCTGTCTCTCTCTGTGTAGGAACCTTggatgttaacttccatgaattcttagtccaagaagcaaaaaag ctgaggtgaaattcagggtgatttaaatgtatcccagatgatgaagactcatatgagattttt tgaacctccagaattcacagtcattggttcacacggaaatgatgatcgttttagaacctgaaagatg aagaggtgagtgctgactatgattaccagaacagactggctgcagcattaggaattaggttcattc cgaacctcatctgaaccaggaaagaaagagttcaattcttactgccttagctctgaggaatgg cactgla/g t tctgttcttcgaacacagataaattgttggttcaaatattcttcccagtc aatattagtaagttcaactgtcaataacctlgcagaacctcagaagcccccttctcaccaacaagc caccacagctgggtcccccactgagacacctcattggcaagactcagttctcaattcttcccacag cagagcatccagcccatattctgaagacccctatagaggatccctTACAGCCAGATGTACACAGG
F5u28	F5	C	T	S	S	cds	GE391	CCCTTCTCTCACCAACAAGC	AGCACGTGAGGCATTTCTGG	CCCTTCTCTCACCAACAAGCcacacagctgggttcccactgagacacctcatlggcaagaactca gttctcaattcttcacacagcagagcatccag c/t ccatattctgaagacccctatagaggatc ctctacagccagatgtcacaggatagcttctacttctacttggctgggagaaattcagaagtc aa gaactgttaagcgaaggaagcccaaggtagaagagatcaagcagcaaaagcacaggttctctctg gatgaattactagcacataaagttgggagacaccttaagccaagacactgggttctctctccggaa tgagccctgggagaccttctctagcaagacactgggttctctctccagaattgagccctgggag gacctctctagtgatctgttactcttaaaacaagtaactcatctcaagatttgggttgggagatg gcatttggcttctggaagaggtagctatgaataatccaaagatactgatgaagacacagctgtta acaattggctgtgacagcccccCAGAAATGCCCTCACCTGCT
F5u29	F5	A	T	K	*	cds	GE389	GAAAGGTAGCTATGAATAAATCAAGA	GGGTCTTGAATGGGGAATGT	GAAAGGTAGCTATGAATAAATCAAGAactgtgatgaagacacagctgttaacaattggctgatca gccccagaatgcctcacgtgcttgggagaaagcacccctcttgcacaagccctggaaagcag agtggccacccaagtttctctagagttagacataaattctctacaagtaagacaggttgaggaaa gagtagactgaagaaagccagtttctcatatgaacacgaaagaaagaaagagagacacacac accatgctctttatctccaggagaccttccacctctaaagagtgaaagcttacaacacatttcca gaaagagacttaagcattcgttgggtgcttcat a/t aatccaatgaacatctcttcccacag acctcaatcagacattgcccctctatggatttgggtggatagcctcacttctgaccataatcag aattcctcaaatgacactgggtcaggaagctgtcctccaggtctttatcagacagtgccccccaga ggaacactatcaaaACATTTCCCATTTCAAGACCC
F5u3	F5	G	A	R	Q	cds	GE47	GAAATACTTTGCAATGAANA	TTTCTGAAAGGTTACTTCAAGG AC	GAAATACTTTGCAATGAANAcaatttctgatatatttcttccaggcagggaacaacaccatga tcagagcagttccaaccagggaacacctatacttataagtggaaacatttagagttttagtaacccc acagaaaaatgatgccagtgcttaacaagaccatactactagctgacgtggacatcatgagagacat cgcctctgggctaataggactacttctaatctgtgaagagcagatccccggacaggc g/a agga atacaggtattttGTCTTGAAGTAACCTTTTCAGAAA

FIG. 5BBB

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
F5u30	F5	C	A	P	T	cds	GE389	GAAGGCTACCT ATGAATAATC CAAGA	GGCTCTTGAAT GGGGAATGT	GAAGGCTAGCTATGAATAATCAAGATactgatgaagacacagctgttaacaattggctgatca gccccggaatgctctacgtgcttggggaagaaagcaccctcttgcacaagcctggaaagcag agtggccaccacaaagtctcttagagtttagacataaatctctacaagttaagacaggtggagaa gagtgaactgaagaaagccagttctctattaaagacacagaaagaaagaaagaaagacacac accatgctctcttctccagagaccttccaccttaagagaggaagcctacaacacatttca gaaagaagacttaagcattcgttgggtcttcaataatcaaatgaacatctctccacagacct caatcagacattgcccctcctatggtatttggctggtatagcctcactt(c/a)ctgaccataatcag aattctcaaatgacactggctcaggcagctgctccaggtcttctatcagacagtgccccaga ggaacactatcaaacattccccattcaagaccc
F5u31	F5	G	A	M	I	cds	GE925	CACTCGCCCTC TCTGTGTC	CAGATTGCCCTT TTCCCTGTATT	CACTCGCCCTCTCTGTGTGTCaacagatttttaattgatttcaactcttngtcnnttcagccattaa tgggatgatctacagcttgcctggcctgaaat(g/a)tatgagcaagagtgggtgaggttacac ctgctgaacataaggcgtctcccaagacattccagtggttcaacttccagggcagaccttgcctgga aaatggcaataaacagacacacagcttaggggtcttgccttctgcctggtaagattgggttaattgg gaaggggccctgcttaagaAATACAGGGAAGAGGCAATCTG
F5u32	F5	T	G	F	V	cds	GE910	GCAGGCAATAT TGGTGTG	TTTACTGATAC AATGCCAGAGT T	GCAGGCAATATTGGTGTGtaattaatcacggaaacacacacatgacaggtgttccacacccctg ggatggaaatggaaagatagaacaacaaacaaatcacagcttctctcg(t/g)taagaaatctt gggtgggagattacttgggaacccctccgtgcccgtctgaatgccagggagcgtggaatgcctgg caagccaaaggctcangtatactctatgcatgggtctctcttagggctccaaagaaagaaagcc ctcaatccacaacacatggaggtcttgggaagcagagatattttaatctgttctgcacaaattaa atcctagggtccaanagtcaggatgcttgggactgatancttgaactataaacttctctcaa aanaagtcattatcatctctgtggttttgggtcttctgatttaaccaaataataagcaaat taatttctgcgaaggatanaatgtaataatttagtcttggggccatacatctctctgcc taactactcaacttGCCATTGTATCAGTAA
F5u33	F5	A	G	D	G	cds	GE921	AAAACCTTAG CCATTATGTT GT	AAGAAGAGAA ATAGTGGAAA C	AAAACCTTAGCCATTATGTTGTCaataaagatttctcttatttgycttccagatttttgaag gaaataactaataacaaaggacatgtgaagaacttctcaaccccccaatcttccaggtttatc cgtgtcattcttaaacatggaatcaaatgacatctgacacttgcctggaaactcttggctgtg(a/g l)atttactagaattgaacattcaaaacccctgggaagagactcttaagaccccaaacattta gaatgggcaatgtattttagctgtgttaaatgttaacagATTTCACATAATTCTCTTCTT
F5u34	F5	A	G	Q	Q	cds	GE932	TCTACTAGAAG TCTGAAGATAT GACAG	TGCCAAATTAC CCATAGAAAT	TCTACTAGAAGCTGAAGATATGACAGactctacttttttgtaatgtgagaaatagatatgaaat atgcttctcaaatagttttttagtatttatttccagtttgaatttctgtaacttctttaaaga aaattgctcagagagatagaaacatttttaagaagaagaaacacaca(a/g)lctaccatttc aggtaagcctgaataatactttttaaattttaaagataaaATTTCATATGGGTAAATTGGCA
F5u35	F5	G	A	A	A	cds	GE362	GCTATCCGAGA TTTGAGACTGG T	TTTGTCCCATG ACAGAACTCC	GCTATCCGAGATTGAGAGTGTGTGaaacacgaatcctcagaacactgccacatgtcttgatggct gttactctccaggtgtcttcttacttgcacacacattccctgc(g/a)gagaagatggacgacg ctgtggctccagggccgagaatacacctatgaaatggagatcagtgagacagtggaacccacct gatgacctccatgcctcacacacatctattactcccatgaaatctgatcgggatttcaactc ggggctgatggggccctgtcttctgtaaaaggtgaagacaccccccccaaaagatccaaca actaaatgttgggaatggctcGGAGTCTGTGTCATGGGACAAA
F5u36	F5	G	A	V	M	cds	GE915	TGATTATCAGA AGAGCAAGGAA A	ACTGTGACCCA GTGTGATTTA	TGATTATCAGAAGAGCAAGAAAtcctgagaagaggcaatacaatttactctgttttccaggg aaaagataatcactcaggttgataggtccctcctaatctgcaaaaggaala-tacataag gacagcaacatgctt(g/a)tggaatgagagaatttcttactatttatgaccttggatgaa agaagagctggtaactatgaagaaagtcgccaggttcttgagactcacatcctcagaaatgaa aaatcccatgagtttcacgggtatttctcctcggactttgatcctaactcctaatTAATTCACACTG GGTCACAGT

FIG. 50CC

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
FF5u37	F5	C	T	P	S	cds	GE391	CCCTTCTCAC CAACAAGC	AGCAGTGAGG CATTCTGG	CCCTTCTCACCAACAGCCaccacagctgggtccc[c/t]cactgagacacctcattggcaagaa ctcagttctcaattctccacagcagagcattccagcccatattctgagagacctatagaggatc ctctacagccagatgtcacagggtacgctactcttctcttctggtctggagaattcagaagtc gaacatgctaagcgttaagggaacccaaggttagaagagatcaagcagcaaacagggtctctctg gatgaattactagcacataaagttgggagacacctaaagcaccgaagacacaggttctctccgga tgaggccctgggaggaccttcttagccaaagacactgggttctctccagaatgagggccctgggag gacctctcagtgatctgttactcttaaaacaagtaactcatctaaagatttgggttgggagatg gcatttggctctgagaaaggtagctatgaataatccaagatactgatgaagcacacagctgtta acaattggctgatcagcccCCAGAAATGCCTCAGTGCT	558
FF5u18	F5	G	C	E	D	cds	GE362	GCTATCCAGA TTTGAGAGTGG T	TTTGTCCTATG ACAGAACTCC	GCTATCCAGATTGAGAGTGGTgaaaacgcaattctcagaacctgccacatgtcttgatggct gttactctccaggtgtcttactctgaccacacattcctctcgga[g/c]aagatggacagc ctgtggctccagggccggaatacacctatgaatggagtatcagtgaggacagtggaacccacct gatgacctctcctcacacacatctattactccctgaatactctgacaggtattcaactc ggggctgattggggccctgcttactgttaaaaggtaagaacaccccccccaaaagattcaaca actaaatgttggaaatgggtcaggagTCTGTCATGGGACAAA	366
FF5u19	F5	G	A	E	K	cds	GE391	CCCTTCTCAC CAACAAGC	AGCAGTGAGG CATTCTGG	CCCTTCTCACCAACAGCCaccacagctgggtccccactgagacacctcatlggcaagaactca gttctcaattctccacagcagagcatccagcccatattctgaagacctatagaggatcctct acagccagatgtcacagggtacgtctacttcttcttctggtctggagaattcagaagtcagaac atgttaagcgttaagggaacccaaggttagaagagatcaagcagcaaacagggtctctctggatg aaattactagcacataaagttgggagacacctaaagcagaacactgggttctctctcgggaatgag gacctgggaggaccttcttagccaaagacactgggttctctccagaatgagggccctgg[g/a]ag gacctctcagtgatctgttactcttaaaacaagtaactcatctaaagatttgggttgggagatg gcatttggctctgagaaaggtagctatgaataatccaagatactgatgaagcacacagctgtta acaattggctgatcagcccCCAGAAATGCCTCAGTGCT	558
FF5u4	F5	T	C	M	T	cds	GE172	TTTAAGAAAT ACAGGTCTCAG CAT	TTTCTCCATG ATTCTGTATTT GT	TTTAAGAAATACAGGTCTCAGCATctggataattctcaaaccaaatggaaacattataaga aagttatgtacacagctacgaagatgagttcttcccaaacatcacagtgaatcccaata[t/c] gaagaagatgggattttgggtctctattatcagagccccagggtcagagacacactcaaatgaatg acacAAATACAGAAATCATGTGGAGAAA	221
FF5u40	F5	C	G	P	A	cds	GE391	CCCTTCTCAC CAACAAGC	AGCAGTGAGG CATTCTGG	CCCTTCTCACCAACAGCCaccacagctgggtccccactgagacacctcatlggcaagaactca gttctcaattctccacagcagagcatccagcccatattctgaagacctatagaggatcctct acagccagatgtcacagggtacgtctacttcttcttctggtctggagaattcagaagtcagaac atgttaagcgttaaggga[c/g]ccaaggtagaagagatacaagcagaagcacaggttctctctg gatgaattactagcacataaagttgggagacacctaaagcagaacactgggttctctccgga tgaggccctgggaggaccttcttagccaaagacactgggttctctctccagaatgagggccctgggag gacctctcagtgatctgttactcttaaaacaagtaactcatctaaagatttgggttgggagatg gcatttggctctlgagaaggttagctatgaataatccaagatactgatgaagcacacagctgtta acaattggctgatcagcccCCAGAAATGCCTCAGTGCT	558
FF5u41	F5	C	G	L	V	cds	GE915	TGATTATCAGA AGAGCAAGGAA A	ACTGTGACCA GTGTGATTTA	TGATTATCAGAAGAGCAAGGAAATccctgagaagaggcaatacaaatctactctgttttccagg aaaagatattcactcggcttgataggtccctc[c/g]taactcgcaaaaagggaata-taca taaggacagcaacatgctatggacatgagagaatttgttactatttattgacctttgatgaaa agaagagctggtactatgaagaagtcgccgaagttcttggagactcacatcccaagaaatga aaatcccatgagttcacgggtatttctctcggactttgactaattctctaatTAATCACACTG GGTCACAGT	334

FIG. 5DDD

Poly Id	Gene	ref Nr	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
F5u42	F5	T	G	Y	D	cds	GE279	GAATTTAGGCCA GTGTGTCACTT G	AGAGATTTCAGA TAGAAATATGC ACAC	GAATTTAGGCAGCTGTGTGACTTGTtgacaaggacagttcttgtttactggctttctctataattgcag gtggacatgcaaaaggagtcataatcacagggtccagagcccaagggtcccaaacattacctgaa gtctgtctatacacagagttc[t/g]atgtagcttacagttccaaccagatcaactggcagatc ttcaagggaacacagcacaaaggaatgtagtggttGTGTGTCATATTTCCTATCTGAATCTCT	255
F5u44	F5	C	A	L	I	cds	GE395	GACCTCAGCCA GACAAACCT	TCTGATCAAGG TCTGGAGGA	GACCTCAGCCAGACAAACCTctctccagaactcagtcagacaacacctttctccagccctcggtcga gatgcccctttctccagacctcagccatacaacctttctctagacttcagccagacaacacctct ctccagaactcagccatagtactctctccagacctcagtcagacaacacctttctccagccctc ggtcagatgcccatttctccagacctcagccatacaacc[c/a]ttctctagacttcagccaga caacctctctccagaactcagtcacaacacctttccccagccctcggtcagatgcccctttct ccagaccagccatacaacacctttctctagacctcagccagacaacacctctctccagaactcag tcagacaacacctttccccagacctcagtcagatgagtgccccttttgcagatctcagtcacaattcccc ttacccagacctcagccagatgacactttctccagacctggtgagacagatctttccccaac tttggtcagatgcccctttccccagacctcagccaggtgactctctctccagacatcagtgacac caccctctccggatctcagccagatatacaccTCTCCAGACCTTGATCAGA	638
F5u45	F5	T	A	L	I	cds	GE923	GATCATTCTCTT TTCCTAGGTT	TTCAGATTACG AGCTTAGGGGA	GATCATTCTTTCTTAGGTTcgttttaaaat[t/a]tagcatccagaccgtattctctacatg cccattggactttctatgaaaaatctatcagagggaagacttatgaagatgactctctgaaatgg tttaaggaaagataatgctgttcagccaaatagcagttatacctacgtatggcatgccactggagc atcagggccagaaagctcctggctcctgctggccttccgcttactactcagctgtgaaacccag taggtactttcattgaaagtttttctcatTCCCTAACCTCGTAATCTGAA	311
F5u46	F5	C	A	L	I	cds	GE387	GTGCCCCCAGA GGAACACTA	TCATATGGCTG AGTCTTGAG	GTGCCCCCAGAGAACACTataaacattccccattcaagaccctgatcaaatgcaactctactctc agaccccggtcacagatctctctccagacctcagtgaaatgcttgagtatgacgaagtcaca agtctctccccagataaagtcaaatgctctccagacctcctcagaacatgaagtcgtgcagacagtc atctctccagacctcagccaggtgacctctctccagaactcagccagacaacacctctctccaga cctcagccacagactctctccagaactcatcagaaaaacctttccccagccctcggtcaga tgccatttctccagacctcagccatacaacacctttctccagacctcagccatacaacacctttct ttagacctcagccagacaacacctctctccagacctcagtcagacaacacctttctccagccctcg tcagatgcccctttctccagacctcagccatacaacc[c/a]ttctctagacttcagccagaca aacctctCTCCAGAACTCAGCCATATGA	548
F5u5	F5	A	G	K	K	cds	GE172	TTTAAGAAAT ACAGGCTCAG CAT	TTTCTCCCATG ATTCTGTATT GT	TTTAAGAAATACAGGCTCAGCACTctggataatttctcaaaccaaatggaaaaacattataaga aagttatgtacacacagtcacgaagatgagctcttcaccaaacatacagtgaaatccaatatgaa a/g]gaagatggattttgggtctctattatcagagcccggtcagagacacactcaagtaagta acAAATACAGAACTCATGGGAGAAA	221
F5u6	F5	G	A	E	E	cds	GE174	TCTATGGTCT GTCTTGTAAC	CAACCACAGGA ATGAAAACTG	TCTATGGCTCTGTTCTTTACCAgactataactgttttctctccagaggcagacacatcgaa cagcaggctgtgtttgtctgtgtttgattgagacaacaaagctggtaaccttgaggacaacatcaacaa gttttgtgaaatctctgtga[g/a]gtgaaactgtgacccccagttttatgatatcaaacatc atgagcagtaagtcagagtagtactattttgttcatCAGTTTTTTCATTCCTGTGGTTG	251
F5u7	F5	C	A	T	T	cds	GE175	TTTACACTTT CAGCTATCAAT GG	GCTTCCTCTGT GAGTGTCCAG	TTTACACTTTTCAGCTATCAATGGctatgtcctcagagacataaactactcttggatctctgcttg atgacactgtccagtgccactctgtagtggtgggagccagaaatgaaatttggaccatccacttc actgggcaactcattcatctatggaagaggtatgagacaccttgac[c/a]ctcttccccatgc gtggagaaatctgtgacggtcacaaatggataatgttggtgtaagagtCTGGACACTTCACAGGG AAGC	264

FIG. 5EE

[illegible]

FIG. 5FF

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')
F7	F7	G	A	R	Q	cds	GE12	CCCTGCAGACC TAGAATGG	CCCATTAACT GCAGAAGATA
F9d8	F9	G	T	-	-	noncoding	GE38	TTCAGATGCGAG AGCATAGAA'TA GA	TGGACTGATCT TTCTGAGTCTCT T
F9u1	F9	G	A	A	T	cds	GE364	GGCCTCAATCT CAATTTTGTA AT	AATAGCCTCAG TCTCCACCT
F9u2	F9	A	T	K	N	cds	GE342	TTTTGCCCTATT CCTGTAAACCAG	GCCCAATTTAT TCAACAACAAAT
F9u3	F9	G	T	R	R	cds	GE364	GGCCTCAATCT CAATTTTGTA AT	AATAGCCTCAG TCTCCACCT
F9u4	F9	A	G	-	-	noncoding	GE303	CAATGAGTATC TACAGGGGAGG A	CACCAATATTG CATTTTCCAAGT

FIG. 511

[illegible]

FIG. 5JJ

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
FGAu10	FGA	G	C	A	P	cds	GE414	GAGGCCTAACA ACCCAGACT	AAAAAGTGTAG TTTCAATGACG TGTA	GAGGCCTAACAACCCAGACTgaggacacatttgaagaggtgtcaggaaatgttaagtcaggagacaa ggagagagtagaccacacagaaactggtcacttctaaaggagataaagagctcaggactcggtaaa gagaaggtcacctctggttagcacaaccacacgctggttctctctctaaaccggttactaagac tgttattggtcctgaggtcacaaagaagtaccacaagaagtggtgacctcgaagatggttctg actgtcccaggagcaatggatttaggcacattgtctgcataggtactctggtggttccgcat aggacacctgatgaa [g/c]ctgctctcttcgacactgcctcaactggaaaacattcccagggt tcttctcactatgttaggagaggttctcagtgagactgagctcagggtcagaaatctggcctc ttcacaaatacaaaaggaaatccagttctcactcaccctgggattagctgaattcccttcccgtgtaa atcttcaagtacagcaaaacaaatttacttagtagcagcaggttacaacagagagactccacattg aaagcaagagctataaaatggcagatgagccggaagtgaagccgcatcatgaaggaaacacatagc accaagagagggccatgctaaatctgcctctgagaggtcagaggtatccacacttctcttgggagacc tccctgtccccttagactaagttaaatatttctgcagaggttcccatggtcccttgcatttcc tcttaactctctgtTACACGTCATGAACTACACTTTT
FGAu11	FGA	G	A	G	E	cds	GE415	TTAACTACCAG GAACTCAATAG ACG	CTGACACCTCT TCAAAATGTC	TTAACTACCAGGAACTCAATAGACGtagtttatgtatttctgtatctacatttctcttatttctc tccctctctctaggtgagacattgatatataagatccgactctgtcagggtcagcagtagggct ttagctcgtgagtagatctgaaggactatgaagatcagcagaagcaacttgaacaggtcattgc caaagacttacttccctctagagataggcaacacttaccactgataaaatgaacacagttccag acttgggtcccggaaattttaaagagccagcttcagaaggttaccctcagagtggaagcattaaac gacatgccgcagatgagaaatggagtttagagagacctgggtgaaatgagattactcaggaggctc cacctctatggaccggatcagagacggaagccccaggaacccctggagctctgggactggagg actcgaacctggaaacctggagctctggacctggagctggaacctggagctggaacctggagctc tggaaactggagctactggaacacaaacctggagctggagccctgagacctggagctggaacctgga atcctggcagctctgaacggaagtgctgggcaactgggcaactggagctctgagctctgta [t/a]ctgg tagtactggacaatggcactctgaatctggaagtttagggcagatagcccaggctctcgggaacg cgaggcctaacaaccagactgggGCACATTTGAAGAGGTCTCAG
FGAu12	FGA	T	A	S	T	cds	GE415	TTAACTACCAG GAACTCAATAG ACG	CTGACACCTCT TCAAAATGTC	TTAACTACCAGGAACTCAATAGACGtagtttatgtatttctgtatctacatttctcttatttctc tccctctctctaggtgagacattgatatataagatccgactctgtcagggtcagcagtagggct ttagctcgtgagtagatctgaaggactatgaagatcagcagaagcaacttgaacaggtcattgc caaagacttacttccctctagagataggcaacacttaccactgataaaatgaacacagttccag acttgggtcccggaaattttaaagagccagcttcagaaggttaccctcagagtggaagcattaaac gacatgccgcagatgagaaatggagtttagagagacctgggtgaaatgagattactcaggaggctc cacctctatggaccggatcagagacggaagccccaggaacccctggagctctgggactggagg actcgaacctggaaacctggagctctggacctggagctggaacctggagctggaacctggagctc tggaaactggagctactggaacacaaacctggagctggagccctgagacctggagctggaacctgga atcctggcagctctgaacggaagtgctgggcaactgggcaactggagctctgagctctgta [t/a]ctgg tagtactggacaatggcactctgaatctggaagtttagggcagatagcccaggctctcgggaacg cgaggcctaacaaccagactgggGCACATTTGAAGAGGTCTCAG

FIG. 5KKK

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
FGAu2	FGA	A	T	G	G	cds	GE415	TTAACTACCAG GAACTCAATAG ACG	CTGACACCTCT TCAAAATGTGC	TTAACTACCAGGAACTCAATAGACCTAGTTTATGATTTGTTATCTACATTTCTCTTTATTTCTTCCCTCTCTAGTGGACATGATATTAAGATCCGATCTTGTGAGGGLCATGCGTAGGGCTTTAGCTCGTAGAGTATCTGAAGGACTATGAAGATCAGCAGAAGCAACTTGAACAGGTCATTGC CAAAGACTTACTTCCCTCTAGAGATAGCAACACTTACCCTGATAAAATGAACAGGTCCTCAG ACTTGGTCCCGG[a/t]AATTAAAGAGCCAGCTCAGAGGTTACCCCTCAGAGTGAAGGCACT AACAGACATGCCAGATGAGAAATGAGTTAGAGAGCCTGGTGGAAATGAGATTACTCGAGGAG GCTCCACCTCTATGGAACCGGATCAGAGACGGAAGCCCCAGGACCTAGCAGTGTGGAAAGC TGGAACCTCTGGAGCTCTGGACCTGGAAAGTACTGGAACCGAAACCTGGAGCTCTGGAGCTGG AGGACTGCAACCTGGAACCTGGAGCTCTGGACCTGGAAGTACTGGAAGCTGGAAGCTCTGGGA GCTCTGGAACCTGGAAGTACTGGAACCGAAACCTGGAGGCTCTGAGCTGCTGAGTACCGGAACC TGGAATCCTGGAGCTCTGGAACCGGAAGTGTGGGACTGGAACCTCTGAGAGCTCTGTATCTGG TAGTACTGGACAATGGCAGCTGATCTGGAAGTTTGGCCAGATAGCCCCAGGCTCTGGGAACG CGAGGCTTAACAACCCAGACTGGGACATTTTGAAGGTGTGAC
FGAu3	FGA	A	T	M	L	cds	GE415	TTAACTACCAG GAACTCAATAG ACG	CTGACACCTCT TCAAAATGTGC	TTAACTACCAGGAACTCAATAGACCTAGTTTATGATTTGTTATCTACATTTCTCTTTATTTCTTCCCTCTCTAGTGGACATGATATTAAGATCCGATCTTGTGAGGGLCATGCGTAGGGCTTTAGCTCGTAGAGTATCTGAAGGACTATGAAGATCAGCAGAAGCAACTTGAACAGGTCATTGC CAAAGACTTACTTCCCTCTAGAGATAGCAACACTTACCCTGATAAA[a/t]TGAAACCACTT CCAGACTTGGTCCCGAAATTTAAGAGCCAGCTCAGAGGTTACCCCTCAGAGTGAAGGCACT AACAGACATGCCAGATGAGAAATGAGTTAGAGAGCCTGGTGGAAATGAGATTACTCGAGGAG GCTCCACCTCTATGGAACCGGATCAGAGACGGAAGCCCCAGGACCTAGCAGTGTGGAAAGC TGGAACCTCTGGAGCTCTGGACCTGGAAAGTACTGGAACCGAAACCTGGAGCTCTGGAGCTGG AGGACTGCAACCTGGAACCTGGAGCTCTGGACCTGGAAGTACTGGAAGCTGGAAGCTCTGGGA GCTCTGGAACCTGGAAGTACTGGAACCGAAACCTGGAGGCTCTGAGTACCGGAACC TGGAATCCTGGAGCTCTGGAACCGGAAGTGTGGGACTGGAACCTCTGAGAGCTCTGTATCTGG TAGTACTGGACAATGGCAGCTGATCTGGAAGTTTGGCCAGATAGCCCCAGGCTCTGGGAACG CGAGGCTTAACAACCCAGACTGGGACATTTTGAAGGTGTGAC
FGAu4	FGA	A	G	T	A	cds	GE415	TTAACTACCAG GAACTCAATAG ACG	CTGACACCTCT TCAAAATGTGC	TTAACTACCAGGAACTCAATAGACCTAGTTTATGATTTGTTATCTACATTTCTCTTTATTTCTTCCCTCTCTAGTGGACATGATATTAAGATCCGATCTTGTGAGGGLCATGCGTAGGGCTTTAGCTCGTAGAGTATCTGAAGGACTATGAAGATCAGCAGAAGCAACTTGAACAGGTCATTGC CAAAGACTTACTTCCCTCTAGAGATAGCAACACTTACCCTGATAAAATGAACAGGTCCTCAG ACTTGGTCCCGAAATTTAAGAGCCAGCTCAGAGGTTACCCCTCAGAGTGAAGGCACTTAAACA GACATGCCAGATGAGAAATGAGTTAGAGAGCCTGGTGGAAATGAGATTACTCGAGGAGGCTC CACCTCTATGGAACCGGATCAGAGACGGAAGCCCCAGGAAACCTGGAGCTCTGGAGCTGGGA ACTCTGGAGCTCTGGACCTGGAAAGTACTGGAACCGAAACCTGGAGCTCTGGAGCTGGAGGG ACTGCAACCTGGAACCTGGAGCTCTGGACCTGGAAGT[a/g]CTGGAAAGCTGGAACCTGGGA GCTCTGGAACCTGGAAGTACTGGAACCGAAACCTGGAGGCTCTGAGCTGCTGAGTACCGGAACC TGGAATCCTGGAGCTCTGGAACCGGAAGTGTGGGACTGGAACCTCTGAGAGCTCTGTATCTGG TAGTACTGGACAATGGCAGCTGATCTGGAAGTTTGGCCAGATAGCCCCAGGCTCTGGGAACG CGAGGCTTAACAACCCAGACTGGGACATTTTGAAGGTGTGAC

FIG. 5LLL

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
FGAu5	FGA	A	G	R	G	cds	GE415	TTAACTACCAG GAACTCAATAG ACG	CTGACACCTCT TCAAAATGTGC	TTAACCTACAGGAACCTCAATAGACGtagtttattgtatttgtatctacattttctctttattttc tccccctctctaggtggacattgataattagatccgatcttgcagggtcagtgtaggagct ttagctcgtgagtagatctgaggaactatgagatcagcagaggaacttgacaggtcattgc caagagacttacttctctctagagataggcaacttaccactgataaaaatgaaacccagttccag acttggttcccgaaatttttagagccagcttcagaggtaccacccagagtggaaggcatataca gacatgccgcagatgagaattgaggttagagagccctggtagaaatgagattactcagggaggtc cacctcttatggaaccggatcagagacggaaayccccaggaaacctagcagtgctggaaagctgga actctggagctctggacctggagctctggacctggaaacctggagctctggagctggaggg actgcaacctggaaacctggagctctggacctggaaagtagctggaaacctggagagctc tggaactggagtagctggaaacccacccctggagccct(a/g)gacctggtagtaccggaaac tggaatcctggcagctctgaatcggaagtgctgggacactctgagagctctgtagctctgtagg tagtactggacaattggcactctgaatctggaaagtttagggcagatagccccaggctctgggaacg cgaggcctaacaacccagactgggGCACATTTGAAGAGGTGTGACG
FGAu6	FGA	C	T	T	T	cds	GE415	TTAACTACCAG GAACTCAATAG ACG	CTGACACCTCT TCAAAATGTGC	TTAACCTACAGGAACCTCAATAGACGtagtttattgtatttgtatctacattttctctttattttc tccccctctctaggtggacattgataattagatccgatcttgcagggtcagtgtaggagct ttagctcgtgagtagatctgaggaactatgagatcagcagaggaacttgacaggtcattgc caagagacttacttctctctagagataggcaacttaccactgataaaaatgaaacccagttccag acttggttcccgaaatttttagagccagcttcagaggtaccacccagagtggaaggcatataca gacatgccgcagatgagaattgaggttagagagccctggtagaaatgagattactcagggaggtc cacctcttatggaaccggatcagagacggaaayccccaggaaacctagcagtgctggaaagctgga actctggagctctggacctggagctctggacctggaaacctggagctctggagctggaggg actgcaacctggaaacctggagctctggacctggaaagtagctggaaacctggagagctc tggaactggagtagctggaaacccacccctggagccct(a/g)gacctggtagtaccggaaac tggaatcctggcagctctgaacgggaagtgctgggacactctgagagctctgtagctctgtagg tagtactggacaattggcactctgaatctggaaagtttagggcagatagccccaggctctgggaacg cgaggcctaacaacccagactgggGCACATTTGAAGAGGTGTGACG
FGAu7	FGA	A	G	K	E	cds	GE414	GAGGCTTAACA ACCCAGACT	AAAAAGTGTAG TTTCAATGACG TGTA	GAGGCTTAACAACCCAGACTggggcacatttgaagaggtgtcaggaaatgtaagtcagggaaca ggagagagtagtaccacacagaaactggtcactctcaaaaggagataaagagctcaggagctggt(a/ g)laagagaaggtccactctggtagcacacacccacgcgtcgttctcgtctcaaaacccgttacta agactgttatttggctctggttcacaaagaaagttaccacaaagagtggtgacctccgagagatggt tctgactgtcccgaggcaattggttttaggcacatttctggtcagataggtactctggtatgggttccg ccataggccacctgatgaagctgctctctcgacactgctcaactggaacacacattccaggtt tcttctcacctatgttaggagagtttgcagtgagactgagtcaggggtcagaatctgggcatc ttcaacaaatacaaaaggaaatccagttctcatcacctgggtagctgaatttcccttccgtggttaa atcttcaagtacagcaacaatttactagtagcacaggttacaacagaggagagactccacattg aaagcaagagctataaaatggcagatgggcccgaagtgaagccgactcatgaagggaacacatagc accaagagaggccatgctaaaatctggccctgtagaggtatccacacitctctcttgggggaagcc tccccgtccccctagactaagttaaatatttctgcagagtggtccccatggcccccttgcatttcc ttcttaactctctgtTACACGTCATTTGAAGAGGTGTGACG

FIG. 5MMM

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
FGAu8	FGA	T	C	P	P	cds	GE414	GAGGCTTAACA ACCCAGACT	AAAAAGTGTAG TTTCAATGACG TGTA	GAGGCTTAACAACCCAGACTggggcacatttgaaaggggtgtcaggaaatgtaagtcaggggacaa ggagagagtagcacacagagaaactggttcactcttaaaaggagataaagagctcaggactggtaaa gagaagggtcacctctggttagcacacacacacgctgcttcactcttaaaacggttactaagac tggtatgggtccctggttagcacaagaagtaccacaggaaggtgtgacctccgaagatgggtctg actgtcccgagggaatggatttaggcacattgtctggcataggtactctggatgggtccgcat aggaccc(t/c)gatgaagctgctcttcgacactgctcaactggaaacacattcccaggtt tcttcacctatgttaggagagttgtcagtgagactgagtcagggggtcagaatctggcatc ttcacaaatacaaaaggaatccagttctcatcaccctgggtagctgaattccctcccggtgtaa atctcaagttacagcaacaaatttactagtagcagcaggttacacagaggagactccacatttg aaagcaagagctataaaatggcagatgagccgggaagtgaagccgacatcagaagacacatagc accaagagaggccatgctaaatctgcccctgtcagaggtalccacactctcctttggggaagcc tcccctgtccccctagactaagttaaatatttctgcacagtggtcccatggcccttgcatttcc ttcttaactctctgtTACACGTCATTGAACCTACACTTTT	821
FGAu9	FGA	G	C	S	T	cds	GE415	TTAACTACCAG GAACTCAATAG ACG	CTGACACCTCT TCAAAATGTGC	TTAACTACCAGGAACCTCAATAGACCTagtttattgtatctacattttctttattttt tcccctctctctaggtggacattgatatgaatccgatcttctgagggttcagtcagtagggct tagctcgtgaagtagattgaaggaactatgaagatcagcagaagcaacttgaaacaggtcatg caagacttacttccctctagagataggcaacacttaccactgataaaatgaacacagttccag actgggtcccggaattttaaaggccagcttcagaaggtaccccgagagtggaaggtcattaca gacatcccgagatgagaatggagttagagagacctggggaatagagattactcgaggaggtc cacctcttatggaccggatcagagacggaaagcccccaggaaaccttagcagtgtcggaaagtga actctggagctctggacctggagtagctggaacccggaacctctggagctctggagctggagg actgcaacctggaaacctggagctctggacctggaaagtactggaal(g/c)ctggaaactctggga gctctggaactggagtagctggaacacaaacccctggagccctgagacctggttagtaccggaacc tggaaacctggagctctggaacggaagtgctgggacactgagacctctgagagctctgtatctgg tagtactggacaatggcactctgaatctggaagtcttaggcccagatagccccaggctctgggaacg cgaggccctaaacacccagactgggGCACATTTGAAGAGGTGTCAG	825
FGBd12	FGB	T	A	-	-	noncoding	GE336	GTAACCATTTT TGAAGTCATTTC CT	CAATTCATT CATACTATAA GCAA	GTAACCATTTTCTGAAGTCATTCTCtagcagagactcagatatataaggattgagatctctcaa tcaagtttaagctctacatgaaaggaatgggttctctggagcttccacaaacttaaaacccatgaaaca tctattatttggctactattgtgtgttttcttagttaagtcctcaaggtgtcacaacgaatgaggagg tgaattttttaaagcattattattattatttagtagtatttattataaagattgaacataatcata ttatgtgcttattttaaataatgaatlagcaTTGCTTATAGTTTATGAATGGAATTG	314
FGBd13	FGB	A	G	K	E	cds	GE336	GTAACCATTTT TGAAGTCATTTC CT	CAATTCATT CATACTATAA GCAA	GTAACCATTTTCTGAAGTCATTCTCtagcagagactcagatatataaggattgagatctctcaa gttaagctctacatgaaaggaatgggttctctggagcttccacaaacttaaaacccatgaaaca tctattatttggctactattgtgtgttttcttagttaagtcctcaaggtgtcacaacgaatgaggagg tgaattttttaaagcattattattattatttagtagtatttattataaagattgaacataatcata ttatgtgcttattttaaataatgaatlagcaTTGCTTATAGTTTATGAATGGAATTG	314
FGBd14	FGB	T	A	-	-	noncoding	GE336	GTAACCATTTT TGAAGTCATTTC CT	CAATTCATT CATACTATAA GCAA	GTAACCATTTTCTGAAGTCATTCTCtagcagagactcagatatataaggattgagatctctcaa gttaagctctacatgaaaggaatgggttctctggagcttccacaaacttaaaacccatgaaacatcta ttattgtctactattgtgtgttttcttagttaagtcctcaaggtgtcacaacgaatgaggaggtaa ttttttaaagcattattata(t/a)ttatttagtattattataaagattgaacataatcata ttatgtgcttattttaaataatgaatlagcaTTGCTTATAGTTTATGAATGGAATTG	314

FIG. 5NNN

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
FGBd15	FGB	C	T	P	L	cds	GE350	TTTCAAAGGTC TATAATAACAC ACTCC	ACACCTGGCCT TGTTTCTCG	TTTCAAAGGTCATATAACACACTCTCTAGTAACTTATGTAATGTTTAAAGAAATGGGTGA CTAAATACAAAGTAATATATGTCATAAACCCCTGAACATATGTTGCTTACATTTGCGAATGTTG AGGAAATATCAGGAAGGAGGTGAACATCTGAATGTTCTTCAACCTGACAGTCTCTGTC AAAC(C/T)GTATAGGTATATCTGTGATGAATGATACAGAAATGAGGTAAAGCTTTCGACAGTTG TTGACCTGTTGATCTGTAATTTATGATACCGTAAATGCAGGAACAAGGCCAGGTGT
FGBd16	FGB	T	C	C	C	cds	GE351	CGCTGAATTTT CCTATGTGCTA TT	TGCATTTCTCGT GGCAGTCTA	CGCTGAATTTTCTTATGTGCTATTTTAAACAAATGTCATGACCCAAATCCTTCACTAATGCTG CTATTTCTTGTGTTTGTAGGGGTGTTG(C/T)CCTACAGGATGTCAGTTGCAAGAGGCTTTG CTACAACAGGAAGGCAATCAGAAATAGTTGTTGATGAGTTAAATAACAATGTGGAAGCTGTTT CCAGACCTCTCTCTTCTTCTGATGATGTTGCTGAAAGACCTGTGGCAAAAGAGGCGAGA AGCAAGTAAAGGTAGATATCTCTGCTTCTTCCATTCGATTTTCAGCTATAAAATGGAAACCGT AGACTGCCACGAGAAATGCA
FGBd17	FGB	T	A	-	-	noncoding	GE392	AAGGAAGAAA GGCAGTTTTT	CCCAGGAAGTG GTAGCTATTAA A	AAGGAAGAAAAGGCGAGTTTCTAGTTTCTCCCAAAATTTTATTTTGGTGAGAGATTTTATTT/a GTTTCTTCTTGTGTAATATGCTTGGCTGGAAATGATAAAATAGCCAGCTTACCAGGATGGGACC CACAGAATTTTGTAGAAATGGAGGCTGGAAAGGAGACAAAGTAAAGGCTCACTATGGAGGAT TCACTGTACAGAAATGAAGCCAAACAAATACCAATCTCAGTGAACAAATACAGAGGAACAGCCGGT AATGCCCTCATGGATGGAGCATCTCAGCTGATGGGAGAAACAGGACCATGACCATTCACAACGG CATGTTCTTCAGCACGTATGACAGAGCAATGACGCGTGGTATGTTGGCACTCTTGTCTCTGC TTTAAAAATCACACTAATATCATTACTCAGATCATTAAACAATATTTTAATAGCTACCACCTTCC TGGG
FGBu1	FGB	C	T	S	S	cds	GE390	TCATAACTGCT TGGTGATAGCT C	CCACTTAGCAT TTTGTGTTGTTG	TCATAACTGCTTGGTGATAGCTCAGTGTAAATAGTTTATCTCAGAAAAATCAAAATGCTAGT TAAATACATTAGTTTATGAGGCAAAATGCTAACTATTTCTACATATTTCTATTTTCCAGATA ATGAAATGTAGTCAATGAGTACTCTCAGAACTGGAAGGAGCAAAATATATATAGATGAGACT GTGAATAG(C/T)AATATCCCACTAACTCTGTTGCTTCTGTTCAATCTTGGAAACCTTGAGAA GCAAAATACAAAGTGAATCTGATGCTCAGCTCAAAATGGAATATGTCGACCCCACTGCACT GTCAAGTGAATATCTCTGTTGCTGGCAAGGTAACTGATCATAACAATATTTTAGAGAG TTCGAGAAGAACTCACACACCAAAATAAGAGAACACAAACACACAAATAATGCTAAGTGG
FGBu10	FGB	G	T	S	I	cds	GE351	CGCTGAATTTT CCTATGTGCTA TT	TGCATTTCTCGT GGCAGTCTA	CGCTGAATTTTCTTATGTGCTATTTTAAACAAATGTCATGACCCAAATCCTTCACTAATGCTG CTATTTCTTGTGTTTGTAGGGGTGTTGCTCCTACAGGATGTCAGTTGCAAGAGGCTTTGCTAC AACAGGAAGGCAATCAGAAATAGTGTGTTGATGAGTTAAATAACAATGTGGAAGCTGTTT CCAGACCTCTCTCTTCTTCTCAGTACATGATTTGCTGAAAGACCTGTGGCAAAAGAGGCGAGA AGCAAGTAAAGGTAGATATCTCTGCTTCTTCCATTCGATTTTCAGCTATAAAATGGAAACCGT AGACTGCCACGAGAAATGCA
FGBu11	FGB	A	G	S	S	cds	GE392	AAGGAAGAAA GGCAGTTTTT	CCCAGGAAGTG GTAGCTATTAA A	AAGGAAGAAAAGGCGAGTTTCTAGTTTCTCCCAAAATTTTATTTTGGTGAGAGATTTTATTTT TCTTTAGGTGAATATGCTTGGCTGAAATGATAAAATAGCCAGCTTACCAGGATGGGACCCACA GAATTTTGTAGTGAATATGGAGGCTGGAAAGGAGCAAAAGTAAAGGCTCACTATGGAGGATTCAC TGTACAGAAATGAAGCCAAACAAATACCAGATCTC(a/g)GTGAACAAATACAGAGGAACAGCGGT AATGCCCTCATGGATGGAGCATCTCAGCTGATGGGAGAAACAGGACCATGACCATTCACAACGG CATGTTCTTTCAGCACGTATGACAGAGCAATGACGCGTGGTATGTTGGCACTCTTGTCTCTGC TTTAAAAATCACACTAATATCATTACTCAGATCATTAAACAATATTTTAATAGCTACCACCTTCC TGGG

FIG. 5000

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
FBu2	FB	A	T	E	V	cds	GE390	TCATTAACCTGCTGGGTGATAGCTC	CCACTTAGCATTTTGTGTTGTG	TCATAACTGCTTGGGTGATAGCTCagtggtttaaataagtttattcttcagaaaaatcaaaatttgtatagttaaatacatattagttttagggcaaaatgctaactattctacataatttccagataatg(a/c)aaatgtatgaatgactcctcagaaactggaagacaaattatataatagatgagactgtgaatagcaatccccaaactctcggtcttcgttcaatccctggaacacctgagaa	452
FBu3	FB	G	T	V	V	cds	GE330	GGGATTCAGATATTATTTCAAACTG	GCTTCCACAA	GGGATTCAGATATTATTTCAAACTGacattatttgcgtgtggttaatatatgctcttttggttctgtcaaccacaaaggatggacagt(g/t)attcagaacacgtcaagacggtagtgtgactttggcaggaaatgggattccataaaacaggatttggaaatgttgaacacacacagatgggaagaattac	289
FBu4	FB	G	A	R	K	cds	GE382	ATGGGTAATCTGCAAAACGTA	TCAAAAAGTCA	ATGGGTAATCTGCAAAACGTAaacttgaccacacgtagttctgttcttaataaacgcaaacacatttctcttcaggttaacatcagatcccagaacacagtgctcaagaagacggtggtggatggtggta	435
FBu5	FB	T	A	G	G	cds	GE477	GAATAGTTACA	TGACTACAGGC	GAATAGTTACATTCCTAATCTTCTAaactctgtatataatttctgcctcatctcttctgtaggg	319
FBu6	FB	A	T	A	A	cds	GE330	GGGATTCAGATATTATTTCAAACTG	GCTTCCACAA	GGGATTCAGATATTATTTCAAACTGacattatttgcgtgtggttaatatgctcttttggttctgtcaaccacaaaggatggacagtgatccagaacggttagttgactttggcaggaa	289
FBu7	FB	A	G	R	R	cds	GE477	GAATAGTTACA	TGACTACAGGC	GAATAGTTACATTCCTAATCTTCTAaactctgtatataatttctgcctcatctcttctgtaggg	319
FBu8	FB	C	T	H	H	cds	GE477	GAATAGTTACA	TGACTACAGGC	GAATAGTTACATTCCTAATCTTCTAaactctgtatataatttctgcctcatctcttctgtaggg	319

FIG. 5PPP

74/178

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
FGBu9	FGB	C	T	Y	Y	cds	GE392	AAGGGAAGAAA GGCAGTTT	CCCAGGAAGTG GTAGCTATTAA A	RAGGGAAGAAAGGCAGTCTTTTtagttcccaaaattttatttttggtagagattttatttttggttt ttcttttaggtgaattattggcttggaaatgataaaatttagccagcttaccaggttggaccacaca gaacttttgatagaaatggaggactggaaaggagacaaagttaaaggctcactatggaggattcac tgtacagaatgaagcccaacaataaccagatctcagtgacaata(c/t)lagaggaaacagcggt aatgcccctcatggatggagatctcagctgatgggagaaacagagaccattcacaaacgg catgtcttcagcacgtatgacagagacaatgacggctggtatgtggcactcttggctcctgc tttaaaatcacactaatatctattactcagatcattacaataattTTTAATAGCTACCACTTCC TGGG
FGGd3	FGG	A	G	M	V	cds	GE337	TTCGTAATAGA CAGCTCTTCAT AGACT	GCAGTTAATT TCTACAAATCA TCC	TTCGTAATAGACAGCTCTTCATAGACTTgacagaggtaaaagattccagaataatgatgtaca tctacagactgttttaggtggcacttactcaaaagcatctactcctaatggttatgataatggca ttatttggggccacttggaaaccccggtggtattccatgaagaaaaccact(a/g)tgagagataat cccatccaacagactcacaaatggagaaggacagcaaacaccctggggggagccaaacaggtca gaccagagcaccctgcggaaacagaaatatgactcactttaccctgaGGATGATTGTAGAAAATT AACTGC
FGGd4	FGG	A	G	-	-	noncoding	GE349	AAATACTTAG CAGTTTCCAAA GAAAA	TGGGTAGCCAC TTTCTAAACTA TTC	AAATACTTAGCAGTCTTCCAAAGAAAAtataaaattactctctctgaaaggaaactatttttgg ctcttatttttggttatcttctgttctgttagataatttgcaggaataataataatcacaata atcaaaagattgttaacctgaagagagtagccagcttgaagcacagtgccaggaaccttgc aaagacacggctgcnaatccatgatatacactgggaaggtaactgatgaaggttatattgggatta ggttcatacaagtaagtaagtgaaggagaaagtgtactgg(a/g)aggtatagGAATAGTTT AGAAAGTGGCTACCCA
FGGd5	FGG	G	A	G	R	cds	GE360	TGCTGATGTGA AAAGTAAGAAA AT	CAAGGTGCTTA GAAAAGTATCT GC	TGCTGATGTGAAGTAAGAAAATtattcttggaaatgaatgaatttactacatgllanaagcta tttttcaaggctggcacagcttacctgcatttcaaacacacagtgaaagtcagttctctctctct agattgtcagaagacttgccaataaggagagtaaacagagc(g/a)ggcttactttttaaacct ctgaagctaacacagcaattcttagtctactgtgaaatcagtggtctggaatggatggactgt gttccagaaggtaatttttccccaccatgtglatttaataaaattcctacattgtttctgcccata tggcagatactTTTCTAGCACCTTG
FGGd6	FGG	T	A	-	-	noncoding	GE372	GAACCAAGTGCT CTGTATTTTGTG AC	CCATTGTCTAT TGATAGTTGGA AAG	GAACCAAGTGCTGTATTTTGTGACaaaatggtgacagcattctctttaca t/a gcattgatag tctattttctctcttcttctctctgcaaatgtgaattagagacttgatggcagtgtagatttcaag aaaaactggattcaataataaagaaggatttggacatctgtctcctactggaacacagaattttg gctgggaaatgagaagatttcttataagaacacagctctgccatcccatatgcatgaagtggtg aactggaagactggaatggcagaaccaggtactgttttgaatgacttccaacttttatttataa agattgcctggaatgtgcaCTTTCCAACTATCAATAGACAATGG
FGGu1	FGG	A	T	Y	F	cds	GE404	CATCCTACGAA ACAGGGAACCTT	TCCACTTCCAG TTTCAAGAAC T	CATCCTACGAAAGAGGGAACCTTctgagatccctgaggagggtcagcatgtgatttcttcc tctctctcagctactgcagact(a/t)tgccatgttcaagggtggagccctgaagctgacaagtaccg cctaacataatgctacttcgctggtgggagatgctggagatgcctttagtggtttgatttggcg atgatccttagtgaacaagtttttccatcccataatggcatgcagttcagtaacctgggacaatgac aatgataagtttgaaggcaactgtgctgaacaggaatggatcgtggtggatgaacaagtgtca cgctggccatctcaatggagtttattaccaaggtatgttttctcttcttagattccaagttaatg tatagtgtatactatttccataaaaaataataaatagatatgaagaaatgaagaaataattataa agatagtaggggattttatcatgttcttcttcttcaactaAGTTCTTTGAAACTGGAAAGTGA

FIG. 5QQQ

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
FGGu2	FGG	A	C	I	I	cds	GE372	GAACCAAGTCTCTGTATTTTG CTGTATTTTGTG AC	CCATTGCTAT TGATAGTTGGA AAG	GAACCAAGTCTCTGTATTTTGACAAaattgttgacagcatctctctacatgcatgatagtctatcttctctcttctgcaaatgtgaattagagacttgatggcagtgtagatttcaagaaaaactggattcaataataaagaaggatttgacatctgtctctacttggcacaacagaatttlygctggaaatgagaagattcatctgat [a/c]agcacacagtctgccatcccatatgcattaaagatggaaactggaagactggaatggcagaaccaggtactgtttgaaatgactccaactttttattgttaaagattgcttggaaatgcaCTTCCCAACTATCAATAGACAATGG	169
FSIRu1	FSIR	A	G	T	A	cds	GE667	GCTCTGAGCTT CATCCAA	AGCAAAAATCC AGCCCATCA	GCTCTGAGCTTCAATCAAAatttgcaacaatatctatttaaggcagaagattgattatattgactcag [a/g]ctagggttcagagatctctctctggcagaagacaatgagtcagctacagcagagatttgacatgacgtgatttgactatgacttatgcaatgaagtgttgactgacctgacttctccctaaagccagatgcattcaacccatgtgaagatatcatgggttacaacatctctcagagctctgatatggttatcagcatctctggccatcactgggaacatcatagtgctagtgtacttaactaccagccaatataaactcacagtcgccaggttctcttatgtgcaacctggcctttgctgactctctgcatggaaatctacctgctcatgcatcagttgatcatccataccaagagccaatatacaaatatgccattgaatggcaaaactggggcaggtgtgtagtctgctggcttttctactgtctttggcagtgagctgtcagctacactctgacagctatcaccttggaaagatggcatcaccatcacgcacgcatgccatgcagctggactgaagggtgcagctccgcatgctgcccagtgctatggTGATGGGCTGGATTTTGGCT	642
FSIRu2	FSIR	G	A	S	N	cds	GE648	GCCATGCTGCC AGTGTC	TGTTTAACTT TGGGCTAAAT	GCCATGCTGCCAGTGTCAatggatgggtggatttttgccttttgcagctgcctcttctcccatcttggcatcagcagctacatgaaggtgagcatctgcctgccccatggatatatgacagcccccttctgctacagctgtatgcatgtccccctcttctgtctcaatgtctctggccttttggctcatctgtggctgctatatccacatctacctcacagtgcggaaccccaacatcgtgtccctctctagtgcacaccagatcggcaagcgcatggccatgctcatcttctactgacttctctgtcatggcaccatctcttctcttctgctcttctgctccctcaagtgccccctcatcagtggttccaaagaaagattctgctgtctgttccaccatcaactctgtgcaaaccttctctatgcccatttaccaaaaacttccgagatgtttcttcatctgtgagcaagtggtggtctgtatgcaatgcaagccccaaatttataggacagaacttcatccactgtccacaacacccatccaaggaatggccactgctctcagctcccaagatcca [g/a]tggttccacttaccatctgtcccctctaaagtcATTAGCCCCAAAACATAAACCA	646
FSIRu3	FSIR	C	G	S	R	cds	GE648	GCCATGCTGCC AGTGTC	TGTTTAACTT TGGGCTAAAT	GCCATGCTGCCAGTGTCAatggatgggtggatttttgccttttgcagctgcctcttctcccatcttggcatcagcagctacatgaaggtgagcatctgcctgccccatggatatatgacag [c/g]cctttgtcacagctgtatgcatgtccccctcttctgtctcaatgtctctggccttttggctcatctgtggctgtcatatataccacatctacctcacagtgcggaaccccaacatcgtgtccctctctagtgcacaccaggtatcgcgaagcgcatggccatgctcatcttcactgacttctctgtcatggcaccatcttcttcttctgcccatttctgctccctcaagggtgccctcatcacgtgttccaaagcaaaagattctgctggttctgttttcaaccccatcaactctctgtgccaccccccttctctatgccatctttaccaaaaacttctgcagagatttcttcatctgtgagcaagtggtggtctgtatgcaatgcaagccccaaatttataggacagaacttcatccactgtccacaacacccatccaaggaatggccactgctctcagctcccaagatcaccagatogttccacttaccatctgttccctctaaagtcATTAGCCCCAAAACATAAACCA	646

FIG. 5RRR

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
FSIRu4	FSHR	T	G	F	V	cds	GE667	GCTCTGAGCTT CATCCAA	AGCAAAAATCC AGCCCATCA	GCTCTGAGCTTCATCCAAAttgcaacaaatctattttaagggaagaagtgattatataagactcag actaggggtcagagatcctctctggcagaagaacaatgagtcagctacagcagagga(t/g)tcg acatgacgtacatgagtttgactatgactatgcaatgaagtgggtgacgtgacctgctccct aagccagatgcatcaacccatgtgaagataatcatggggtacaacatcctcagagtcctgatatg gtttatcagcatcctggccatcactgggaacatcatagtgctagtgatccttaactaccagccaat ataaactcacagtcaccagggttcttattgtgcaacctggccttctgctgctctgcatggaaatc tacctgctgctcattgcatcagttgatatccataccaagagccaatatacacaactatgccattga ctggcaaaactggggaaggtgctgctgctggccttttctactgtcttggccagtgaagctgtcag tctacactctgacagctatcaccttggaaagatggcataccatcacgcatgccatgcagctggac tgcaagggtgcagctccgcatgctgcccagtgctcattggTGATGGCTGGATTTTGGCT	642
FSIRu5	FSHR	G	A	T	T	cds	GE667	GCTCTGAGCTT CATCCAA	AGCAAAAATCC AGCCCATCA	GCTCTGAGCTTCATCCAAAttgcaacaaatctattttaagggaagaagtgattatataagactcag actaggggtcagagatcctctctggcagaagaacaatgagtcagctacagcagaggaatttgacat gac(g/a)taactgagtttgactatgactatgcaatgaagtgggtgacgtgacctgctccct aagccagatgcatcaacccatgtgaagataatcatggggtacaacatcctcagagtcctgatatg gtttatcagcatcctggccatcactgggaacatcatagtgctagtgatccttaactaccagccaat ataaactcacagtcaccagggttcttattgtgcaacctggccttctgctgctctgcatggaaatc tacctgctgctcattgcatcagttgatatccataccaagagccaatatacacaactatgccattga ctggcaaaactggggaaggtgctgctgctggccttttctactgtcttggccagtgaagctgtcag tctacactctgacagctatcaccttggaaagatggcataccatcacgcatgccatgcagctggac tgcaagggtgcagctccgcatgctgcccagtgctcattggTGATGGCTGGATTTTGGCT	642
FSIRu6	FSHR	A	C	I	L	cds	GE667	GCTCTGAGCTT CATCCAA	AGCAAAAATCC AGCCCATCA	GCTCTGAGCTTCATCCAAAttgcaacaaatctattttaagggaagaagtgattatataagactcag actaggggtcagagatcctctctggcagaagaacaatgagtcagctacagcagaggaatttgacat gacgtacactgagtttgactatgactatgcaatgaagtgggtgacgtgacctgctcccttaagc cagatgcatcaacccatgtgaagataatcatggggtacaacatcctcagagtcctgatatggttt atcagcatcctggcc(a/c)tcactgggaacatcatagtgctagtgatccttaactaccagccaat ataaactcacagtcaccagggttcttattgtgcaacctggccttctgctgctctgcatggaaatc tacctgctgctcattgcatcagttgatatccataccaagagccaatatacacaactatgccattga ctggcaaaactggggaaggtgctgctgctggccttttctactgtcttggccagtgaagctgtcag tctacactctgacagctatcaccttggaaagatggcataccatcacgcatgccatgcagctggac tgcaagggtgcagctccgcatgctgcccagtgctcattggTGATGGCTGGATTTTGGCT	642
FSIRu7	FSHR	A	G	T	A	cds	GE667	GCTCTGAGCTT CATCCAA	AGCAAAAATCC AGCCCATCA	GCTCTGAGCTTCATCCAAAttgcaacaaatctattttaagggaagaagtgattatataagactcag actaggggtcagagatcctctctggcagaagaacaatgagtcagctacagcagaggaatttgacat gacgtacactgagtttgactatgactatgcaatgaagtgggtgacgtgacctgctcccttaagc cagatgcatcaacccatgtgaagataatcatggggtacaacatcctcagagtcctgatatggttt atcagcatcctggcc(a/g)ctgggaacatcatagtgctagtgatccttaactaccagccaat ataaactcacagtcaccagggttcttattgtgcaacctggccttctgctgctctgcatggaaatc tacctgctgctcattgcatcagttgatatccataccaagagccaatatacacaactatgccattga ctggcaaaactggggaaggtgctgctgctggccttttctactgtcttggccagtgaagctgtcag tctacactctgacagctatcaccttggaaagatggcataccatcacgcatgccatgcagctggac tgcaagggtgcagctccgcatgctgcccagtgctcattggTGATGGCTGGATTTTGGCT	642
FSHu1	FSH	T	C	Y	Y	cds	GE611	GGAACCTCCAC AATACCATAAC CTA	TTATCTTTCA TTTCCACCAAG G	GGAACTTCCAAATACCATAACCTAactctctctttaaactcctcaggatctggtgataaaggac ccagccaggccccaaaaatccagaaaaacatgaccttcaagggaactggta(a/t/c)gaacagtgga gagtggccgggtgctgctcaccatgcagatcctcttataataacccagtggtgccccacccagtgtaac tgtggcaagtgtagcagcagcagcactgattgtactgtggaggccttggggccacgactactgctc CTTTGGTGAATGAAGATAA	282

FIG. 5SS

[illegible]

FIG. 5TTT

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
GH1u1	GH1	G	A	-	-	noncoding	GE527	AAGGGCCAGG GTATAAAA	TCTCCCTCAG GACACATTG	AAGGGCCAGGGTATAAAAAGggcccaagagacc(g/a)gctcaaggatcccaagggcccaact cccgaaccactcagggtccctgtggacagctcacctagctgcaatggctacaggtgaagcggccct aaaaatcccttggggcaaatgtgtctTGAGGGGAGA	166
GH1u2	GH1	A	C	-	-	noncoding	GE527	AAGGGCCAGG GTATAAAA	TCTCCCTCAG GACACATTG	AAGGGCCAGGGTATAAAAAGggcccaagagaccggctcaaggatcccaagggcccaactcccc g(a/c)accactcagggtccctgtggacagctcacctagctgcaatggctacaggtgaagcggccct aaaaatcccttggggcaaatgtgtctTGAGGGGAGA	166
GH1u3	GH1	T	G	-	-	noncoding	GE527	AAGGGCCAGG GTATAAAA	TCTCCCTCAG GACACATTG	AAGGGCCAGGGTATAAAAAGggcccaagagaccggctcaaggatcccaagggcccaactcccc gaaccactcagggtccctgtggacagctcacctagctgcaatggctacaggtgaagcggccct aaaaatcccttggggcaaatgtgtctTGAGGGGAGA	166
GH1u4	GH1	C	G	S	C	cds	GE609	GGGGGAGACCT GTAGTCAG	GTCAGTGGGC TCCAAGATT	GGGGGAGACCTGTAGTCAGAGccccggggcagcagggcccaatggccctccccgcagaac ctgagagctgctccgcatct(c/g)ccctgctctcaccagctcggtggcggcccgctgagctccc tcaggagtgcttccggcaacagcctgggtgacggccctctgacagcaacgctctatgacctccta aaggacctagaggaaggcatcccaacgctgagtggggtggggtggcgtagggtccccAAATCT TGAGGCCCACTGAC	275
GH1u5	GH1	A	T	-	-	noncoding	GE527	AAGGGCCAGG GTATAAAA	TCTCCCTCAG GACACATTG	AAGGGCCAGGGTATAAAAAGggcccaagagaccggctc(a/t)aggatcccaagggcccaact cccgaaccactcagggtccctgtggacagctcacctagctgcaatggctacaggtgaagcggccct aaaaatcccttggggcaaatgtgtctTGAGGGGAGA	166
GH1u6	GH1	G	C	-	-	noncoding	GE527	AAGGGCCAGG GTATAAAA	TCTCCCTCAG GACACATTG	AAGGGCCAGGGTATAAAAAGggcccaagagaccggctc(a/g)atcccaagggcccaact cccgaaccactcagggtccctgtggacagctcacctagctgcaatggctacaggtgaagcggccct aaaaatcccttggggcaaatgtgtctTGAGGGGAGA	166
GH1a9	GH1	C	G	-	-	noncoding	GE602	GCTACAACATG ATTTTGGAAAC A	GCTTCCCATTT TATTTAGTCT	GCTACAACATGATTTTGGAAACAAatcaatcttttttaacccttcatctttaggaacactcaagaa tggactcaagaaatggaaagaatgcctgattatgtttctctgctgggaaacacagctgttactttaa ttcatcgtttacctccatctggataaccttalljlatcaagctcaactagcaatgggtggacagtggt atgaaaagtgttctctgttgatgaaataggLaatcacaggtttttgttttcatcttgacatagtt t(c/g)AGACTAAATAAATGGGGAGC	287
GH1d7	GH1	G	A	R	H	cds	GE597	TTGAGTTGTG ACTCTTTGGC	TGACAAAGCC AGGTTAGC	TTGAGTTGTGACTCTTTGGCCaatatggcgtttatattttgtcttgaaagatggacctatat tgaacaacatcagttccagtgtaactcattgaaagtggataaggaatatgaagtc(g/a)tgtag atccaaacaaacgaacacclttggaaattatggcaggttcagtgaggtgctctatgtaaacactcctc agatgagccaatttaccatgtgaagaaggtaaaagaataaaagataaaatagtaGCTAACCTGG CTTTGTCA	269
GH1d8	GH1	C	T	-	-	noncoding	GE597	TTGAGTTGTG ACTCTTTGGC	TGACAAAGCC AGGTTAGC	TTGAGTTGTGACTCTTTGGC(c/t)aatatggcgtttatattttgtcttgaaagatggacct atatttgacaacatcagttccagtgtaactcattgaaagtggataaggaatatgaagtgcgtgtgag atccaaacaaacgaacactctggaaattatggcaggttcagtgaggtgctctatgtaaacactcctc agatgagccaatttaccatgtgaagaaggtaaaagaataaaagataaaatagtaGCTAACCTGG CTTTGTCA	269
GH1u1	GH1	A	G	G	G	cds	GE596	TTAAATTTGTCT GTCTTGTA CT	GAAAGAAAGT CAAAGTGAAG G	TTAAATTTGTCTGTCTTGTAaattgctctgttgtaattgcaagtgcaaccagatccaccat tggcctcaactggactttactgaacgtcagtttaactgggattcattgagatcccaagtgagat gggaaagcaccacgcaatgcagataattcagaagg(a/g)tggaagggtcttgagatgaacttca atacaagaagtaaatgaaactaaatggaaatggtaagatgttgctacaCCTTACACTTGGACT TTCTTTC	268

FIG. 5VVV

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
GHRu2	GHR	A	C	I	L	cds	GE649	GCCCATATTCA GCTAAGCAAT	ATTGCCCCAGT CAATTCTTT	GCCCATATTTCAGCTAAGCAATCCaagttccactgtcacaacatcgacttttatgccagggtgagcga cattacaccagcaggtagtggctcttcccgggcacaagaataggcagggtgagtgcccaat gtgacatgcacccggaaatggctcactctgccagaacacttcttatggacaatgctacttc tgtgaggcagatgccaaaaagtgc[a/c]tccctgtggtcctcacatcaaggttgaatcacaca tacagccaagcttaaaccaagaggacattatcacaccacagaaagccttacactgctgtg999 aggcctgggacaggagaacatgttccagggttctgagatgctgtcccagactatacctccattca tatagtacagtccccacaggcctcatactaatgcgactgcttgccttgcctgacaagaggt ttctctcatcatgtggtctatgtgagcagacacacactgaacaaaaatcatgaccttgcctttt ggtttcccaagagctacgtatttaattagcAAAGAAATTGACTGGGGCAAT
GHRu3	GHR	G	A	R	II	cds	GE596	TTAAATTGTGT CTGCTCTGTGA CT	GAAAGAAAGT CAAAGTGTAAAG G	TTAAATTGTGTCTGTGTACTAatgctctgttgaattgcacagtgcaaccagatccaccat tgccctcaactggactttactgaacgtcagtttaactggatttcacagatatccaagtggat gggaagcaccac[g/a]caatgcagatatcagaagaggtggatgggttctggaagtatgaacttca atacaagaagtaaatgaactaaatggaaaatggtaaggttgctacacacCTTACACTTTGACT TTTCTTTC
GHRu4	GHR	G	T	C	F	cds	GE1207	GCCATTCATGA TAGCTATAAAC C	CTCACCTGGGC ATAAAAGT	GCCATTCATGATAGCTATATAACCCgaattccacagtgatgactcttgggttgaatttatgagct agatatgtgagccagatgaagaagactgaggaaatcagacacagacagacttcttaagcagtgacc atgagaaatcacatagtaacctagggtggaaggtggcagctctggacgtaccagctgttgtgaa cctgacattctggagactgatttcaatggcaatgacatatactgaggtacctcagaggttgcctca ggcacagaggttaaaagggaagcagatctcttatgctttaccagagaagaatcaaaataaactcac cttatcatgattgtt[g/t]cctctactcagcagccagtggttatccaaagcagaggaaaaaaca accacaaccacttctactgaaggagctgactcaactcacaagctgcccataatcagctaaagca atccaagttcacgtcacaacatcgACTTTTATGCCAGGTGAG
GHRu5	GHR	C	A	P	T	cds	GE649	GCCCATATTCA GCTAAGCAAT	ATTGCCCCAGT CAATTCTTT	GCCCATATTTCAGCTAAGCAATCCaagttccactgtcacaacatcgacttttatgccagggtgagcga cattaca[c/a]cagcaggtagtggtgctcttcccgggcacaagaataggcagggtgagtgctc caatgtgacatgcacccggaaatggctcactctgccagaacaaacttcttatggacaatgctactc cttctgtgagcagatgccaaaaagtgatccctgtgctcctcacatcaatcaaggttgaatcacaca tacagccaagcttaaaccaagaggacatttatacaccacacagaaagccttacactgctgtg999 aggcctgggacaggagaacatgttccagggttctgagatgctgtcccagactatacctccattca tatagtacagtcaccaagggcctcactactcaatgcgactgcttgccttgcctgacaagaggt ttctctcatcatgtggtctatgtgagcagacacacacactgaacaaaaatcatgaccttgcctttt ggtttcccaagagctacgtatttaattagcAAAGAAATTGACTGGGGCAAT
GHRu6	GHR	C	A	P	T	cds	GE649	GCCCATATTCA GCTAAGCAAT	ATTGCCCCAGT CAATTCTTT	GCCCATATTTCAGCTAAGCAATCCaagttccactgtcacaacatcgacttttatgccagggtgagcga cattacaccagcaggtagtggtgctcttcccgggcacaagaataggcagggtgagtgctc gtgacatgcacccggaaatggctcactctgccagaacaaacttcttatggacaatgctacttc tgtgaggcagatgccaaaaagtgatccctgtgctcctcacatcaaggttgaatcacacataca gccaaagcttaaaccaagaggacatttatacaccacagaaagccttacactgctgtg999ggg c/a]ctgggacaggagaacatgttccagggttctgagatgctgtcccagactatacctccattca tatagtacagtcaccaagggcctcactcaatgcgactgcttgccttgcctgacaagaggt ttctctcatcatgtggtctatgtgagcagacacacacacactgaacaaaaatcatgaccttgcctttt ggtttcccaagagctacgtatttaattagcAAAGAAATTGACTGGGGCAAT
GHRu7	GHR	G	C	W	S	cds	GE599	TCAACCTGTCT TGGATCTAATT T	GGGGTATCCT GAATGTTTAAT A	TCAACCTGTCTGTCTAATTgatttgccatttcattgctcctagaatgaagccaattcaaaa ctcctagctggccttattctactgactt[g/c]gtggtggaagggtgctccagccagcactggc cctatggactgcgcccggagaaagagagatgccgaaaatttgattgatctcttcccaagaggt agtttctctcagcttcaaaataagacatagttgatttcaatttaactatATTAAACATTTCAG GATAGCCCC

FIG. 5WWW

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
GNRHRu 2	GNRHR	C	T	F	F	cds	GE569	GGAAACACCA 'TTTCATTCT'	CAAGGCTTAT CCACCTCTA	GGAAACACCATTTCAITTTCTTatctccatctcaagcatcacattctcttcttcagatagtc aaagaggttggtcaactggcagaaacccaagctt c/t gaatgcaccacgaccagccagctt ctccccccgagacctgaagagctctgtaagttaaagtgatcataacatgacacagcatag agctctAGAGGTGATAGCCTTG
GNRHRu 3	GNRHR	A	C	-	-	noncoding	GE578	CATTAAAGGC 'TTTATGTGAGG AT	TCTAAAGAAGA AAACTCGTGC TA	CATTAAAGGCTTTATGTGAGGATTTTaaatattaccattaaaaaaagcatagtcatt tgcatataattaccagcaggaagatttcaatgtcctggaaaaaattccctataaaaaaggaaga taggaacacagaaagtcacagtaactcaactactcaaggaagat.tgggatcttttggctct ctggctctaaacaggttaa a/c aggtcttgattatttctTACACGAGTTTCTTTTAGA
GNRHRu 4	GNRHR	C	A	S	Y	cds	GE599	TCAACCTTGTC 'TGGATCTAATT 'I'	GGGGCTATCCT GAATGTTTAAT A	TCAACCTTGCTGGATCTAAITTTgatttgctgattcatgtgcttgaatgaagccaattcaaaa ctcclagctggccttattctactgacttggtgctggaagctgct c/a cayccagcactgggt cctatggactgcccctggaggaagagagatgccgaatttggatcttcttccaagagga agttctctcagcttcaaaaataagacatagtgatttgattcaatttaactataTTAAACATTCAG GATAGCCCC
GNRHRu 5	GNRHR	G	T	-	-	noncoding	GE599	TCAACCTTGTC 'TGGATCTAATT T	GGGGCTATCCT GAATGTTTAAT A	TCAACCTTGCTGGATCTAAITTTgatttgctgattcatgtgcttgaatgaagccaattcaaaa ctcctagctggccttattctactgacttggtgctggaagctgctccagcagcactggctcta tggaactgcccctggaggaagagagatgccgaatttggatcttctccaagag g/t ta agttctctcagcttcaaaaataagacatagtgatttgattcaatttaactataTTAAACATTCAG GATAGCCCC
GPIBAu 2	GPIBA	C	T	T	M	cds	GE493	CTTCCAGGGGA 'TGCAGG	AAAAGCAAAAG GCAGGAGGT	CTTCCAGGGGATGCAGGGgagatccactcaagggtcccttgcccaaggtctcctctctc ctcttgctgctcctgctgccaagcccttacacccccaccccatctgtgaggtctccaaagtggc cagccacctagaagtgaactgtgacagagaatctgacagcgtgctccagacacctgccgaag acacacacatctccactgagtgagaaacctctctgacaccttccctggcaacctgatgct tacactgctcactcagctgaacctagataggtgcagctcaccagctccaggtcgatgggac gctgcaagtgtgggacctggactatcccaaatcagctgcaagcctgccccttgcctagggc agacactgctgctcactgctcctggagctctcttcaacggctgacctgctgctcttgggt gcccgtggtcttgccgaactccaagagctctacctgaaggcaatgagctgaagacctgccc cccagggtcctga c/t tgcccaacacacagctggagagctgagctggttaacacaaacttg actgagctccccgctgggtcctgaatgggtggagaaatctcgacacctctcctccaagagaa ctcgtgtatacaataccaaagggtcttcttgggtccccACCTCTGCTTTTCTTT
GPIBAu 3	GPIBA	C	T	N	N	cds	GE495	GACACCTTCT CCTCCAAGA	GATTGGGGTGG GCTCCG	GACACCTTCTCTCCACAGaaactcgctgtatacaataccaaagggtcttcttgggtccacct cctgcttctgcttctccacgggaacctctggttatgcaactgtgagatctctcttctgctc gctggtcgcaggacaatgctgaataatgtctacgtatggaagcaaggtgtggacgtcaaggccatg acctctaa c/t gtggccagtgtgagtgagacaaatcagacaaagtctccgtctacaaatacc caggaaggggtgccccaccttggtgtagaagtgacacagacctatgattactaccagaa gaggacactgagggcgataaggtgctgccaagagactgtggtcaagtccccaccaaaggccca tacaacccccctgggtctctactctgctgctcactgctctctagacagccaatgacctcct ccttgcatccaacacagaatccactaaggagcagacacattcccacctagatggaccccaaat ttcacacttcacatggatccatcacattctccaaactccaaatccactactgaaccaacccc aagcccgaccacctcagagccgtccccggagcccccccaaacatgaccacctggagccactc caagcccgaccacccagagccacctcagagccccccagccccgagccccCGGAGGCCACCC CCAATC

FIG. 5XXX

FIG. 5YYY

[illegible]

[illegible]

FIG. 5AAAA

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
GRIN1u1	GRIN1	G	C	E	Q	cds	GE1287	GAGCTGAGAGG AGACTGCCG	GTAGTGCGGG CGGTAAC	GAGCTGAGAGGAGACTGCCGccctgggcagccttagtgcgtgggtccaggctgggtctcccttc ccccagatttgacagatccaccag[g/c]agccttcgtgtacgtcaagcccaagctgagtgag tgggacatgcaaggaggagtccacagtcaacggcaccacagcaagaagtgatctgcaccgggc ccaacacacgtcgccgggcagccgtgagtcgcggggcaggcgcggggcgcggggcaggcgcc ggggcggtggggcggtctggagccagcagctTACCGCCCGCACCTAC
GRIN1u5	GRIN1	T	G	C	G	cds	GE1287	GAGCTGAGAGG AGACTGCCG	GTAGTGCGGG CGGTAAC	GAGCTGAGAGGAGACTGCCGccctgggcagccttagtgcgtgggtccaggctgggtctcccttc ccccagatttgacagatccaccaggaaccccttcgtgtacgtcaagcccaagctgagtgag aca[t/g]gcaaggaggaggtccacagtcacggcagccacagtcacaagaagtgatctgcaccgggc ccaacgacacgtcgccgggcagccgtgagtcgcggggcaggcgcggggcgcggggcaggcgcc ggggcggtggggcggtctggagccagcagctTACCGCCCGCACCTAC
GRIN1u6	GRIN1	G	C	D	H	cds	GE1287	GAGCTGAGAGG AGACTGCCG	GTAGTGCGGG CGGTAAC	GAGCTGAGAGGAGACTGCCGccctgggcagccttagtgcgtgggtccaggctgggtctcccttc ccccagatttgacagatccaccaggaaccccttcgtgtacgtcaagcccaagctgagtgag acatgcaaggaggaggtccacagtcacggcagccacagcaagaagtgatctgcaccgggccaa c[g/c]acacgtcgccgggcagccgtgagtcgcggggcaggcgcggggcgcggggcaggcgcc ggggcggtggggcggtctggagccagcagctTACCGCCCGCACCTAC
GRIN1u7	GRIN1	G	A	K	K	cds	GE1115	GCGGAGCTGG GAGGAC	AGGGACGGAG GTCAGC	GCGGAGCTGGGAGGACGctgctgcacgccccgcggtctgtgcgctcgccaggtgaaacaacagc aaacaaggaggagtggaatggatggatggcgagctgctcagcgggcaggcagacatgactcgtggc ggcgtaccataaacaacgagcgcgcgagtcacagatcagagtttccaaagccctcaagtaccagg gcctgactattctggtcaa[g/a]aagtgggcgaggcggtgtggcggttggcggggggga gtccctggaggggcccgggcgccgctGACCTGCGGTCCCT
GRIN1u8	GRIN1	G	A	E	E	cds	GE1120	TTCCGGCAGTG GGAGGC	CGTCTGCACC TCGGCT	TTCCGGCAGTGGGAGCGgggtggagggcggttccccgggttccacctcagccacagtgcccc cgctcccgagagcaccacctgagcttctgcgcacgtgcgcgctactccccacagtcacag cgtgtggtttgagatgagtcgtgtctacagctggaaacacatcctctgctggtcagcgacgacc acga[g/a]ggccggcggtcagaacgcctggagacgctgctggaggagcgtgagtcacaggt gagggtcgggcgcggggtggcgccctggcgaggcagcagctGCGAGCTGCAGGACG
GRIN1u17	GRL	G	T	-	-	noncoding	GE1196	ATTCAAGGTGG CCAAATTAT	AAAGAAACAA AAACATGTCC	ATTCAAGTGGCCAAATTATttgtgtaagaaactgaaaaatctaatattaaaaaatatggaact tctaataattttatatttagttatagtttcagatatataatcatatggtatctcaataatctgg gaagggaagggtcactcagcttccatgcgaatttataaaaatgattgtaaaatagcttgtatag tgtaaaataagaatgatttttagatgagattgtttatcatgacatgttataattttttgtagg [g/t]gtcaagaaatgctgagtaacctatagttatagtttgcacatgcatatcatalcatcacag gcagcaggtgtctcagaacacacagtttgccttaggggaagaggagatggagagactgctctg tgtgcagtgaaaggttgcgtgagctctgaccagtgagtcgtcagcgaggttttagtttactcaatctc catctgacaccccttctcatcccaacagtgagtcgtcagcgaggttttagtttactcaatctc cccttgactaaagtatgtaaagtatgaacagagagcagccctccccagcaggttgaatgacaacagaa ggcaccatctaatagcgggttacttccatacacagcctccccagcaggttgaatgacagaaatctca gcttcagaagtttggcaatagtttgcatagaggtaccagcaatatgtaaatagtcagaaatctca taggttgcccaataatcactaatctcttctatcctacacagaggtttattttccaaataaagt aGGACATGTTTTGTTTTCTTT
GRIN1u18	GRL	G	T	-	-	noncoding	GE128	GACCTTCCCAT TACAGTN.ATT TC	GAAGAAACAC AAAGGTTTATA TAGTTGC	GACCTTCCCATTACAGTTCATTTCtatgtattt[g/t]tttaaataccacagctcgaaaaacaa agaaaaaataaaaggaaatccagaggccactacaggagcttcacaagaaacccctgtaaaatcct ggttaacaaacaaatagttctctgacaggttaccacaaactacccctacccctggtgtcactgttggaa ggttatgaaacctgaagtgttatgcaaggatagatagctctgttccagactcaacttggaggaa tcatgactcagctcaacatgttagggggcggaagtgattgcaagctgaaatgggcaaggca ataccaggtgaagatgcaaaacataaaagagcCAACTATATAAACCTTTGTTTTCTTC

FIG. 5BBB

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
GRLd19	GRL	G	A	E	E	cds	GE666	TGTGACTTTAG AGCTTATGATG TT	GACCCAGAAGA AAACTCCAAA	TGTGACTTTAGAGCTTATGATGTTTcccccggttttttggtttttttggtttttggtagtggatattcac tgatggactccaaagaatcatctaactcctggttagaagaagaaacccacagcagtgctgctgtctcag ga[g/a]aggggagatgtgatggactctataaaacctaaagagggagctactgtgaagggtt ctgctctccacctcaactggctgtcgcttctcaatcagactccaagcagcgaagacttttggtt gattttccaaaaggctcagtaagcaatgcgcagcagccagatctgtccaaagcagtttcaactctc aatgggactgtatatgggagagacagaaacaaagtgatggaaatgacctgggattccacacagc agggccaaatcagccttctcggggaacacagactaaagcttttgggaagaagcatcgcaaac ctcaataggctcgaccagtggtccagagaaaccccaagagttcagcatccactgctgtgtctgtgc ccccacagagaaggagttccaaaactcactctgatgtatcttcagaaacagcaaatgtgaagg gccagactggcaccacacggtggcaatgtgaattgtataccacagacccaagcacctttgacatt ttgcaggatttggagtttcttctgggtc
GRLd20	GRL	G	A	R	K	cds	GE666	TGTGACTTTAG AGCTTATGATG TT	GACCCAGAAGA AAACTCCAAA	TGTGACTTTAGAGCTTATGATGTTTcccccggttttttggtttttttggtagtggatattcac tgatggactccaaagaatcatctaactcctggttagaagaagaaacccacagcagtgctgctgtcag gaga[g/a]gggagatgtgatggactctataaaacctaaagagggagctactgtgaagggtt ctgctctccacctcaactggctgtcgcttctcaatcagactccaagcagcgaagacttttggtt gattttccaaaaggctcagtaagcaatgcgcagcagccagatctgtccaaagcagtttcaactctc aatgggactgtatatgggagagacagaaacaaagtgatggaaatgacctgggattccacacagc agggccaaatcagccttctcggggaacacagactaaagcttttgggaagaagcatcgcaaac ctcaataggctcgaccagtggtccagagaaaccccaagagttcagcatccactgctgtgtctgtgc ccccacagagaaggagttccaaaactcactctgatgtatcttcagaaacagcaaatgtgaagg gccagactggcaccacacggtggcaatgtgaattgtataccacagacccaagcacctttgacatt ttgcaggatttggagtttcttctgggtc
GRLu1	GRL	T	G	F	V	cds	GE666	TGTGACTTTAG AGCTTATGATG TT	GACCCAGAAGA AAACTCCAAA	TGTGACTTTAGAGCTTATGATGTTTcccccggttttttggtttttttggtagtggatattcac tgatggactccaaagaatcatctaactcctggttagaagaagaaacccacagcagtgctgctgtcag gagagggagatgtgatggactctataaaacctaaagagggagctactgtgaagggttctgc gtctccacctcaactggctgtcgcttctcaatcagactccaagcagcgaagacttttggttgat t/g)ttccaaaaggctcagtaagcaatgcgcagcagccagatctgtccaaagcagtttcaactctc aatgggactgtatatgggagagacagaaacaaagtgatggaaatgacctgggattccacacagc agggccaaatcagccttctcggggaacacagactaaagcttttgggaagaagcatcgcaaac ctcaataggctcgaccagtggtccagagaaaccccaagagttcagcatccactgctgtgtctgtgc ccccacagagaaggagttccaaaactcactctgatgtatcttcagaaacagcaaatgtgaagg gccagactggcaccacacggtggcaatgtgaattgtataccacagacccaagcacctttgacatt ttgcaggatttggagtttcttctgggtc
GRLu10	GRL	A	G	-	-	noncoding	GE196	ATTCAAGGTGG CCAAATTAT	AAAGAAACAA AAACATGTCC	ATTCAAGGTGGCCAAATTATTTgtgtaatagaaacactgaaatctaataattaaaaatatgggaact tctaataatttttataattagttatagtttcagataataatacatattggtattcactaatactgg gaagggaagggtactcagctttacatgcaatttataaaatgattgtaaaaatagcttgtatag tgtaaaaataagaatgatttttagatggatgttttatacatgacatgttatatatattttgttagg ggtaaaagaaatgctgtgataaacctatatgatttttagtttgtacatgacattcatalcagcagcag cgatgggtctcagaaaccaaacagtttgcctcagggaagagggagatggagactggctctgtgtg cagtgaaaggttgcgtgaggtctgacccagtgcagattacagaggaagttatcctctgcctccatt ctgaccaccttctcattccaaagtgatctgcagcaggttttagtttaactaatctccct tgactaaaagtatglaaaglatglaaacaggagacaggaaggtgggtgcttacatccttaaggca ccatctaataagcgggttactttcacatacagccctccacagcagttgaatgacaacagagctt cagaagtttggcaatagtttgcatagaggtaccagcaatatgataatagtcagaaatctcatagg ttgccataataacactaatctcttctt[a/g]tctacacaaagagtttatttccaaataaaatg AGCACATTTTGTGTTTCTTT

FIG. 5CCC

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
GRU11	GRL	A	C	-	-	noncoding	GE558	AAATATGTTTG AAGACCTGTGA A	AAATGATAAAT TTTTATAAGCT ACAGAG	AAATATGTTTGAACACCTGTGAAACCTTAAATAGTGCCTTTATCCATAT [a/c]ggacagcaca at tacctatgtgcggaaggaaatgatgcacatcgataaaaattcgaagaaaactgccagca tgccgtatcgaaaatgtctcaggctggaatgaacctggaaggtaaatataaatatctgaaagca attgcttctCTCTGTAGCTTATAAANAATTTATCAATT
GRU12	GRL	C	T	H	H	cds	GE565	CTTCTGAAGAG TGTTGCCCTCAT	GGGAAATGAC ACACATACAAC T	CTTCTGAAGAGTGTGCCCTCATACCTTTATCTCTAAATCAGGTTTCAGGAACCTACA [c/t] ctggatgaccaaataagccctactcagcagctactcctggatgttctcttatggcatttgcctctggggctg gagatcatatagacaatcaagtgcacacctgctgtgttttgcctcctgatctgatatttaaatgagt aAGTTCTATGTGTGCATTTTCCC
GRU13	GRL	T	A	-	-	noncoding	GE1325	TGATCTGTCAA ACTTCCAGAAC C	TTGGCATTGCT GTAAATG	TGATCTGTCAAACCTTCCAGAACCATGGTAGCCTTCAGTGAGATTCCCATCTTGGCTGGTCACTCC ctgactgtagctgtagtggaatgtgttttggctgtgtgtctgtgttttagtgcagaaggaa ataaaagctgaaggaggaacatt [t/a]aaaccttggggtaggttctgtaatttcccagacta tttccaagcaacctggtccaccaggattagtgaccagggttttcaggaaggatttgcctctctc tagaaaatgctcgaaggattttatttctgtatgaaaggctgtatgaaatccccctcccaata acttgcctaaactacatatagattcaagtggtgtcgaatattctatttcttatataaatgctatata atggggacaaatctatatattactgtgtatggcattataagaagcttttccattatttttctc acagtaattttaaattgttaaaaataaaaaccagtgctcctgtttaaataaaaagtgtagt ttttattcattgctgaataataatctgtagtttaaaaaaaaagtgtcttttaccacagcagtgaa atgtcagactgtaaaaaccttgtgtggaatgtttaaactttatttttccatttaaatgtctgtt ctggattaccacacacacatttgcacgaattggcagtaaatgttagcCAITTTACAGCAATGC CAA
GRU14	GRL	G	C	D	H	cds	GE1201	CAACGGTGSCA ATGTGAAA	AAGAAACAGGA AAACACTGAT	CAACGGTGGCAATGTGAAATgtataccacagaccaaagcaccttttgacattttgcaggaatttgg agttttctcttgggtccccaggtaaaagagacgaatgagagtccttggagatcagacctgttgata gatgaaaaactgtttgcttctctctcctcggggagaaagacgattcattctcttttggaaggaaactc gaatgaggactgcgaagcctctcatttaccggcacataaccacaaaataaggataaatggagatc tggtttgtcaagccccagtaattgaacactgcccaagtgaaaacagaaaagaagatttcatc gaactctgcaacctggggttaattaaagcaagagaaaactgggcacagtttactgtcaggcgaagctt tcctggagcaaatataatggtaataaaatgtctgccatttctgtctatctgtgtgtgagtaacctctg gaggacagatgtaccactatgacatgaatacagcatcccttttccaacagcag [g/c]atcagaa gcctatttttaatgtcatccacaaattcccgttgggtccgaaaattggaatagggtgccaaaggat ctggagatgacaaactgacttctctgggactctgaactccctcctggtcgaacagtttttctaat ggctattcaagggttaagATCAGTGTGTTTTCCTGTTCTT
GRU15	GRL	C	A	-	-	noncoding	GE564	GCATTTTTCAT TTATGCAATGG	GAGGAATTAAT TTGTCTGATTA AAA	GCATTTTTCATTTATGCATGGaaacctgaaaaaagtttacaagtgatatacagaaaagggaagtl tgtgccttttatagctattactgtctgggttttaacaatttccctttatatttagtgaactacgctt gtctcattttttctacataattttttattcaagttattgtacagctgtttaagatgggcagctag ttcgtagctttcccaataaaactcctaaacatttaactcaatcatctgtgtgaaaaatttgggtt tcaacctgatggcacttagctatcagagacacaaaattgactcaaatctccagatttcttctg tcaaaaaaaataaaagctcatttttgtatataatgtcttctcagtggaataatataggt tgtgcaaatataacagtcctaa [c/a]tggtatagacacactagccagtgacctgtctgggttaaac tgtggaatggttgcaaaagactaaattaaaaataataaccaagagccccgtctgtactaaac cgccctatttttgcaatggctatatggcaagaaagctgttaaacatttcttctcaggacacctt tgaagtagtttgtataacttcttaaaagttgtgtatccagataaccagctgtatacacagctgaga gaCTTTTAAATCAGACAAAAGTAATTCCTC

FIG. 5DDDD

FIG. 5EEFF

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')
GRLu5	GRL	T	C	-	-	noncoding	GE671	TGATCCAGAT AACCCAGCTGTAC	GCATTTCCTTG ACCCCTACAA
GRLu6	GRL	A	G	-	-	noncoding	GE1325	TGATCTGTCAA ACTTCCAGAACC	TTGGCATTGGCT GTAAATG
GRLu7	GRL	A	G	P	P	cds	GE666	TGTGACTTTTAG AGCTTATGATG TT	GACCCAGAAGA AAACTCCAAA
GRLu8	GRL	C	T	P	P	cds	GE1201	CAACGGTGGCA ATGTGAAA	AAGAAACAGGA AAAACACTGAT

FIG. 5FFFF

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')
GRLu9	GRL	G	A	-	-	noncoding	GE664	GCATTTTTCGATTGATGCTAATGG	GAGGAATTACTTGTCTGATTA AAA
HSD3B1 d1	HSD3B1	C	T	V	V	cds	GE593	GCCAGATACAGAAATCATTTCCA A	TAATTCCTCCACCTTGCTGC
HSD3B1 d2	HSD3B1	A	G	I	V	cds	GE593	GCCAGATACAGAAATCATTTCCA A	TAATTCCTCCACCTTGCTGC
HSD3B1 d3	HSD3B1	T	A	G	G	cds	GE593	GCCAGATACAGAAATCATTTCCA A	TAATTCCTCCACCTTGCTGC
HSD3B1 d4	HSD3B1	C	T	L	L	cds	GE668	GAGCTTCCTTAC TCAGGCCCAA	GGAGAAAGGAG GAAAGTATGTA T
HSD3B1 d5	HSD3B1	G	T	-	-	noncoding	GE668	GAGCTTCCTTAC TCAGGCCCAA	GGAGAAAGGAG GAAAGTATGTA T

FIG. 5GGG

91/178

Assay Sequence										
Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	
HSD3B1 d6	HSD3B 1	T	C	F	L	cds	GE675	GGTGGCACCTCTAGGGGATATATCTGACAGTGAACAATATGCTCTCATGGACAGGLACCAGC	GGCGGCTCGAT AGGTGTAAA	692
HSD3B2 d25	HSD3B 2	G	A	A	A	cds	GE1194	CAGAAGAATGC ACCCTGAGTC	GCCAGATCTCG CTGAGCC	839
HSD3B2 d26	HSD3B 2	A	G	T	T	cds	GE1194	CAGAAGAATGC ACCCTGAGTC	GCCAGATCTCG CTGAGCC	839
HSD3B2 d27	HSD3B 2	C	T	-	-	noncoding	GE639	AAAATAAGGCA TCTGCTGAGTC TAT	CCATGCAGAGT TTAAGATGGAG	338

FIG. 5HHHH

Assay Sequence									
Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')
U3D3B2 028	U3D3B2 2	C	G	-	-	noncoding	GE665	CCTGCTGGAAATAGTGAGCTTC	GCTCTTTTGTGTAAGTGTGTA
627									
CCTGCTGGAAATAGTGAGCTTCctactcagcccaatttacacctatcaaccccccttcaaccgcccacacagtcacattatcaaatagcgtattcacctctcttacaagaagctcagcgagatctggcgataaagccactctacagctggaggaagcccaagcagaacacgtggagtggttgggttcccttgtggaccggcacaagagagaccctgaagtcgaagactcagtgatttaaggatgacagagatgtgcattggttattgttaggaattgcatcaaaactccaccacctggcctcatcacagaagcaacaaggccacaagcccaggtctgctgctcccttccacaaatgcccacttactgtattcctcatgtcatcacaagcccaggtcactggcccaacaagaacgtttctgtcctaatacatcacccagaagaaaaaataatgattgctgttaccaaatctcagtagctgattctgaacaattgaggaccctttaaactgaagggcccttttgactaatagagctccatttccacttc/g)ttaatgagaaagcatttcccttctctttaaattctccatttccctTCACACAGTTCACAAAGAGC									
U3D3B2 029	U3D3B2 2	C	T	-	-	noncoding	GE665	CCTGCTGGAAATAGTGAGCTTC	GCTCTTTTGTGTAAGTGTGTA
627									
CCTGCTGGAAATAGTGAGCTTCctactcagcccaatttacacctatcaaccccccttcaaccgcccacacagtcacattatcaaatagcgtattcacctctcttacaagaagctcagcgagatctggcgataaagccactctacagctggaggaagcccaagcagaacacgtggagtggttgggttcccttgtggaccggcacaagagagaccctgaagtcgaagactcagtgatttaaggatgacagagatgtgcattggttattgttaggaattgcatcaaaactccaccacctggcctcatcacagaagcaacaaggccacaagcccaggtctgctgctcccttccacaaatgcccacttactgtattcctcatgtcatcacaagcccaggtcactggcccaacaagaacgtttctgtcctaatacatcacccagaagaaaaaataatgattgctgttaccaaatctcagtagctgattctgaacaattgaggaccctttaaactgaagggcccttttgactaatagagctccatttccacttc/g)ttaatgagaaagcatttcccttctctttaaattctccatttccctTCACACAGTTCACAAAGAGC									
U3D3B2 01	U3D3B2 2	C	G	R	R	cds	GE639	AAAATAGGCATCTGCTGAGTGAT	CCATGACAGGT
338									
AAAATAGGCATCTGCTGAGTGATaaccattttacctcttcttttagccctcttctgggtcacgctagaatcagatctgctctccagcatctctgtttctctggcaagtgttctctgacttctgggttgccacgatgacgggtggagctgctgtgtgacaggaaggggtcttctgggtcagagatcttgcccg/c/g)ctgtgtgtggaggaagaaactgaaggagatcaggcccttgacaaggccctcagaccagaattgagagaggaaatttcttagtaagtaaaacttgagtcattgggtctgtggtCTCCATCTTAACTCTGCATGG									
U3D3B2 010	U3D3B2 2	A	G	-	-	noncoding	GE665	CCTGCTGGAAATAGTGAGCTTC	GCTCTTTTGTGTAAGTGTGTA
627									
CCTGCTGGAAATAGTGAGCTTCctactcagcccaatttacacctatcaaccccccttcaaccgcccacacagtcacattatcaaatagcgtattcacctctcttacaagaagctcagcgagatctggcgataaagccactctacagctggaggaagcccaagcagaacacgtggagtggttgggttcccttgtggaccggcacaagagagaccctgaagtcgaagactcagtgatttaaggatgacagagatgtgcattggttattgttaggaattgcatcaaaactccaccacctggcctcatcacagaagcaacaaggccacaagcccaggtctgctgctcccttccacaaatgcccacttactgtattcctcatgtcatcacaagcccaggtcactggcccaacaagaacgtttctgtcctaatacatcacccagaagaaaaaataatgattgctgttaccaaatctcagtagctgattctgaacaattgaggaccctttaaactgaagggcccttttgactaatagagctccatttccacttc/g)ctgattctgaacaattgaggaccctttaaactgatttcttgaactaatagagctccatttccactctttaaattgagaaagcatttcccttctctttaaattctccatttccctTCACACAGTTCACAAAGAGC									

FIG. 5III

[illegible]

FIG. 5JJJJ

FIG. 5K KKK

[illegible]

FIG. 5L1LL

FIG. 5MMMM

[illegible]

FIG. 5NNNN

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
HSD3B2 u9	HSD3B 2	T	C	-	-	noncoding	GE665	CCTGCTGGAAA TAGTGAGCTTC	GCTCTTTTGT TGAACGTGTGTG AA	CCTGCTGGAAATAGTGAGCTTCctactcagcccaatttacacctatcaaccccccttcaacggcc acacagtcacattatcaaatagcgtattcaccttctcttaagaaggctcagcgagatcttggcg tataagccactctacagctggaggagagcccaagcagagaaaaacccgtggagtggttggctcccttgt ggaccggcacaaagagacccctgaagctccaagactcagttatttaaggatgacagagatgtgcacg tgggtattgttaggaaatgtcatcaaatccacccacctggctcatagagaaagcaacaagggc acaagcccaggctcctgctgctccctttcaccaatgcccacttactgtattctcatgtcatc aaaacctgcacagtcactggcccacaagaacgtttctgtcctaatacatcaccaagagaaaaac aataatgattgtc/c/gttaccaaatctcagtagctgattctgaacaattgagggaaccccttaa actgaaggggccctttgactaatagagctccatttccactctttaaatgagaaagcatttccctttc tctttaattctccatttctTCACACAGTTCAACAAAAGAGC
HTR1ad 9	HTR1A 9	G	A	V	V	cds	GE1181	GACTGTTTGCT AGTGGGGAGA	TGAGGAAGCCA ATAAGCCAA	GACTGTTTGCTAGTGGGGAGACTccagctccggcagccagttcggagcgggcaaatgga cagcgacagacagagcttccagccacctctcccgccgggagatcctggagctgttccaggcc aactccagtttccagctggagcttctgaacgcttggaacgctggagagccagggagcgccctgaa agctgctcctcggaataaccttcgccaagcagtaagaacttctgttgggtctctgcatctcc cttctccgaaacttccagagagagggcggaagaccacccaggggaaggggcgaggaatcttcg cgctgttttcttctccctccctcccgccggcgccgagggcaggtggaatgtgctcagccctgg tcagggaacaacacacatcaccacccgctccctttgagaccggcggaacactactggtatct ccgagctgacgctcagctaccaagtgtacacctctctgtgtgggacgctccttcagaaagtgccaa gtgctgggcaatgctggtggtggctgctccttcgagcgtcctggagcgtccttcagaaagtgccaa ttatcttattggctcttggcggtcacgacctcatggtgtcgtgtgtgtgctgccccatggccg cgctgtatcaggt(g/a)ctcaacaagtggaacactggcgccaggttaacctgacgtgttcatcgc cctcgagctgctgtgctgacctcatcttgcacctgtgcgacctgagcaggtgacgtgacgt gggccatcacggaccctcagactacgtgaacaagagggagccccggcgccgctgctgctcctc tcgctcacTTGGCTTATTGGCTTCTCTCA
HTR1AU 1	HTR1A 1	C	T	A	V	cds	GE1181	GACTGTTTGCT AGTGGGGAGA	TGAGGAAGCCA ATAAGCCAA	GACTGTTTGCTAGTGGGGAGACTccagctccggcagccagttcggagcgggcaaatgga cagcgacagacagagcttccagccacctctcccgccgggagatcctggagctgttccaggcc aactccagtttccagctggagcttctgaacgcttggaacgctggagagccagggagcgccctgaa agctgctcctcggaataaccttcgccaagcagtaagaacttctgttgggtctctgcatctcc cttctccgaaacttccagagagagggcggaagaccacccaggggaaggggcgaggaatcttcg cgctgttttcttccctcccttcccgccggcgccgagggcaggtggaatgtgctcagccctgg tcagggaacaacacacatcaccacccgctccctttgagaccggcggaacactactggtatct ccgagctgacgctcagctaccaagtgtacacctctctgtgtgggacgctccttcagaaagtgcc /t)gggtgctgggcaatgctgctggtggtgctgctccttcgagcgtccttcagaaagtgcc ccaatattcttattggctcttggcggtcacgacctcatggtgtcggtgtgtgtgctgccccatg gccgctgtatcaggtgtcacaagtggaacactggggccaggttaacctgcgacctgtcatcgc cctcgacgtgctgtgctgacctcatcttgcacctgtgcgacctcgcgctggagcaggtact gggccatcacggaccctcagactacgtgaacaagagggagccccggcgccgctgctgctcctc tcgctcacTTGGCTTATTGGCTTCTCTCA

FIG. 50000

FIG. 5PPPP

[illegible]

FIG. 5QQQQ

[illegible]

FIG. 5RRRR

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
HTR1DB u3	HTR1D B	C	T	S	S	cds	GE1163	CTTCGTCCGCTCCATGCCC	CACACAGCATT CCGACA	CTTCGTCCGCTCCATGCCCaaagagctgcgctccggagctggggcgagagagccatggaggaaacc 661 gggtgctcagtcgctccaccgcccgcgggctccgagacctgggtctctcaagccaacttat cctctgctccctcccaaaactgacgcccgaaggactacatttaccaggactc c t atctccct acctggaaaagtactgctgttatgctattggcgctcatcacttggccaccgctctccaatg cctttgtgattggccacagtgaccggaccggaacacaccccggttaactacctgacgccc tctctggggltcaaccgacctgcttgggtccatcctggtgatgcccacagaccatgtacactgt cacggccgctgggacactgggcccagggtgctctgtgacttctggctgctgctcggaacatcacttgtt gcactgcctccatcctgcacctctgtgctcatcgccctggaccgctactggggccatcacggacgccc gtggagtactcagctaaaaggactcccaaggggcggtcatgacgcgctggtgtgggtctt ctccatctctatctcgtcgccgctctctctggcgctcaggctaaaggccgaaggaggTGTCCG AATGCGTGGTG
HTR1DB u4	HTR1D B	T	G	V	G	cds	GE1163	CTTCGTCCGCTCCATGCCC	CACACAGCATT CCGACA	CTTCGTCCGCTCCATGCCCaaagagctgcgctccggagctggggcgagagagccatggaggaaacc 661 gggtgctcagtcgctccaccgcccgcgggctccgagacctgggtctctcaagccaacttat cctctgctccctcccaaaactgacgcccgaaggactacatttaccaggactcctcctacc tggaaaagtactgctgttatgctattggcgctcatcacttggccaccgctctccaalgcctt tgtgattggccacagtgaccggaccggaacacaccccggttaactacctgacgctctc tggcggtcacgacctgcttgtgtccatcctggtgatgcccacagaccatgtacactgtcac ggccgctggacactgggcccagggtg t g ctgtgacttctgggtgctcggaacatcacttgtt gcactgcctccatcctgcacctctgtgctcatcgccctggaccgctactggggccatcacggacgccc gtggagtactcagctaaaaggactcccaaggggcggtcatgacgcgctggtgtgggtctt ctccatctctatctcgtcgccgctctctctggcgctcaggctaaaggccgaaggaggTGTCCG AATGCGTGGTG
HTR1Du 1	HTR1D	G	C	V	L	cds	GE1162	TTGAAGGAAGG AGCCAAATG	TGPAGGAGATC TGAGAGGTGTT CA	TTGAAGGAAGGAGCAAAATGtgagggtctgtggaggtctgtgggaagagagaccactagcatgtccccact 650 gaaccagtcagcagaaggccttccccaggaggctcccaagatccttgatgacacagaaacct cagaggcttgggtccaggaacctccaggcgctcaagatctccttggcgtggctcttccgctc atcacactggccca g c tctctccaatgctt t tactacacacacacttactcaccagga agctccacacccctgccaactacactgattggctccttggccaccgacctcttgggttccatc tgglaatgcccacatcagcatcgctataaccatccacacacctggaaacttggccaaatcttgtg tgacatctgggtgctctctgacatcacgctgctgcacagcctccatcctgcatctctgtgtcattg ccttggacaggtactgggcaatacacagatgcccctgggaatacacgtaaacgagagcgtggccac ggggccacacatgacgcctatgtctgggacatctccatctgcatctccatcccccgctctctg ggggcaggcccaaggccagaggagatgtcgggactgtctgtgTGAACACCTCTCTCAGATCTCTTACA
HTR1Du 10	HTR1D	C	T	S	L	cds	GE1161	AGGAGGAGATG TCGGACTGT	ATCCCGATGAG GTTACAGGA	AGGAGGAGATGTCGAGCTGTctgtgtgaacacctctcagatctctctacacacatctactccacctgt 648 ggggcccttctacatctccctcgtgtgtgtctcatcctctatattggccggatctacccggctgccc gaaccgcatcctgaatccacctcactctatgggaagcgttccacagggccacccatcacaag gctctgcccgggtcctcgtctgctcgctcaactccagcctccatgaggggcact c t gcactc ggctggctccccctctctt t tcaaccacgtgaaaatcaagcttgcagacagtgcctgggaacgca agaggatttctgctgctcgagaagaaagccactaaaatcctgggcatcattctgggggcccctt atcatctgctggtgccccttctctgtgtgtctgtgtgctccctccatctgcccggactcctgctg gatccacccggcgtctt t gacttctt cacttctcaggtggtat t aaactccctcacaatccaa taatctacactgtgtt t aatgaaggtt t cggcaagctt t tcagaaaat t g ccccct t ccggaag gctcctctagctct t t cgtgtgacgcgcaagaaact t t gtgTCTGTAACTCATCTCGGAT

FIG. 5SSSS

FIG. 5TTTT

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')
HTR1D ₅	HTR1D	A	G	T	T	cds	GE1162	TTGAAGGAAGG AGCCAAATG	TGTAGGAGATC TGAGAGGTGTT CA
HTR1D ₆	HTR1D	T	C	I	I	cds	GE1162	TTGAAGGAAGG AGCCAAATG	TGTAGGAGATC TGAGAGGTGTT CA
HTR1D ₇	HTR1D	C	G	S	S	cds	GE1162	TTGAAGGAAGG AGCCAAATG	TGTAGGAGATC TGAGAGGTGTT CA
HTR1D ₈	HTR1D	A	C	A	A	cds	GE1162	TTGAAGGAAGG AGCCAAATG	TGTAGGAGATC TGAGAGGTGTT CA

FIG. 5UUUU

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
HTRIEL u1	HTRIEL	C	T	I	I	cds	GE1162	TTGAAGGAAGG AGCCAAATG	TGTAGGAGATC TGAGAGGTGTT CA	TTGAAGGAAGGAGCCAAATGctggagggtctgtgggaagagagagagccacccatgcatgtcccccact gaaccagtcagcagaaggccctcccccaggagccctccacagagatccctgaatgccacagaaacct cagaggcttggggtatccaggaccctccaggcgctcaagatctcccttggcgtggctcttccgtc atcacactggccacagtcctctcaatgcttctgactcaccacatcttactcaccaggaagct ccacacccctgccaatcactgattggctccctggccaccacccaccccttgggttccatcttggg taatggccatcagcatcgctataccatcaccacacccctggaaacttggccaaatcttgtgtgac atctggctcctctgacatcacgtgctgacagctccatccatctctgcatctctgtgtcatgtctct ggacaggtaactgggcaatcacagatgcccctgggaatacagtaaacgagagagagctggccacggg ccacatgctgcccattgtctgggcccattccatctgcatctccatcccccgtctcttggcg g/t]caggccaaaggccaggagagatgtcggaactgtctggTGACACCTCTCAGATCTCTTACA
HTRIEL u2	HTRIEL	C	T	I	I	cds	GE1160	TTCCCTTGTTA CAGGTATCCAT	GTGGGATGTAG AAAGCTCCA	TTCCCTTGTTACAGGTATCCATtttccagctatatataatcttttaaaacaaagaaaatggatttc ttaaatctcatctgatcaaaacttgacctcagaggaactgttaaacagaaatgccatccaaatctt gggtccctcactctgtctgggtggcactgagacaaactatcaactcccttggat[c/t] gctgcaatttatgtgacccgggaagctgcacatccagccaatatttaattgttcccttgcagt cacagatttcttggctgctgtgacatctggctgaggttggacttaccattacgttgcagtgctcc ggattatgggcaagtggtctgtgacatctggctgaggttggacttaccattacgttgcagtgctcc atcttgcatctcagctatagcttggatcggtatcgagcaatcacagatgctgttgagatgc caggaaaaggactccaaagcatgctggcatatgattacaatagtttggattatatctgtttta tctctatgctcctctcttcttgaggccacaaaggaactgagagatgagaaatgcatcatcaag cacgacacacattgttccacacatttactcaacatttggagcttTGTATATCCAC
HTRIEL u3	HTRIEL	T	C	I	T	cds	GE1160	TTCCCTTGTTA CAGGTATCCAT	GTGGGATGTAG AAAGCTCCA	TTCCCTTGTTACAGGTATCCATtttccagctatatataatcttttaaaacaaagaaaatggatttc ttaaatctcatctgatcaaaacttgacctcagaggaactgttaaacagaaatgccatccaaatctt gggtccctcactctgtctgggtggcactgagacaaactatcaactcccttggatcgtg caatta[t/c]tgtagcccggaagctgcacatccagccaatatttaattgttcccttgcagt cacagatttcttggctgctgtgacatctggctcagcttgcgtatgtgtatgtgagagagagct ggattatgggcaagtggtctgtgacatctggctgaggttggacttaccattacgttgcagtgctcc atcttgcatctcagctatagcttggatcggtatcgagcaatcacagatgctgttgagatgc caggaaaaggactccaaagcatgctggcatatgattacaatagtttggattatatctgtttta tctctatgctcctctcttcttgaggccacaaaggaactgagagatgagaaatgcatcatcaag cacgacacacattgttccacacatttactcaacatttggagcttTGTATATCCAC
HTRIEL u3	HTRIEL	T	G	V	G	cds	GE1160	TTCCCTTGTTA CAGGTATCCAT	GTGGGATGTAG AAAGCTCCA	TTCCCTTGTTACAGGTATCCATtttccagctatatataatcttttaaaacaaagaaaatggatttc ttaaatctcatctgatcaaaacttgacctcagaggaactgttaaacagaaatgccatccaaatctt gggtccctcactctgtctgggtggcactgagacaaactatcaactcccttggatcgtg caatttatgtgacccgggaagctgcacatccagccaatatttaattgttcccttgcagtcaca gatttcttgggtgctgctgggtgacatgcccctcagcttggatattgtgagagagagctggat tatggggcaagtgg[t/g]ctgtgacatttggctgaggttggacttaccattacgttgcagtgctcc atcttgcatctcagctatagcttggatcggtatcgagcaatcacagatgctgttgagatgc caggaaaaggactccaaagcatgctggcatatgattacaatagtttggattatatctgtttta tctctatgctcctctcttcttgaggccacaaaggaactgagagatgagaaatgcatcatcaag cacgacacacattgttccacacatttactcaacatttggagcttTGTATATCCAC

FIG. 5VVVV

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
HTR1EL u4	HTR1E L	A	T	E	V	cds	GE1160	TTCCTTGTGTTA CAGGTATCCAT	GTGGATGTAG AAAGTCCA	TTCCCTTGTACAGGTATCCATTTTtcagctatatatttaaaacaaagaaalggatttc tcaaatctcatctgatcaaaacttgacctcagagaaactgttaaacagaatgccatccaaatlct gggtccctcactctgtctggctggcactgatgaacaactatcaactcccttgatcgctg caattattgtgacccggaagctgcacccatccagcaattatttaattgttcccttgagtcaca gatttcttctgtgctgtcctggatgaccttcagcatgtgatatgtgagagagagctggat tatggggcaagtgtctgtgacatttggctgagtggtgacattacctgctgcagtgctccatct tgatctctcagctatagcttggatcggtatcgagcaatcacagatgctgtg[e/l]glatgc caggaaaaggactccaaagcatgctggcatttatgattacaatagttggatttatctgttttta tctctatgctctctctatctggaggcaccaaaggactagcagagatggtgaatgcacatcaag cagaccacattgttccacatttactcaacatttggagcttTGAGGCTTTTCTACATCCAC	640
HTR1EL u5	HTR1E L	T	A	I	I	cds	GE1158	CGACCACATTG TTTCCACCA	AGTTATTCCTC CCCTCAAAA	CGACCACATTGTTTCCACCACTtactcaacatttggagcttctctacatcccactggcattgatt tgatctcttactacaataatataagagcagcaagacattataccacaagagacaagcaagtagg attgcaaaaggaggtgaatggcgaagtccttctggagagtggtgagaagaaagcactaaatcagt ttccacatctctatgtactagaagaagcttcttctgacccatcaacagacttctgataaat[t/a] catagcacagtgaagctctcaggctcgaattcgaagcatgagaaatcttggagaagcacaagat ctcaggtacaagacggaagcagccactacccctggatttaattcttgggtgcatcttgaatat gttggcttctcttcttctglaaaagaaatagttgttaattctctgacaaatglaaaattctgaa gaaatgtccaatcttctggcatggcttgggtatctcaaatctccctataaatccactgatttacac aatctttaaagacttcaagaagcatcccaagacttgcgaatgtgcgatttagtttttaa aatgtttattattgaaggatgggggtTTTGGGGGAGGAATAACT	631
HTR1EL u6	HTR1E L	T	G	D	E	cds	GE1160	TTCCTTGTGTTA CAGGTATCCAT	GTGGATGTAG AAAGTCCA	TTCCCTTGTACAGGTATCCATTTtctcagctatatatttaaaacaaagaaalggatttc tcaaatctcatctgatcaaaacttgacctcagagaaactgttaaacagaatgccatccaaatlct gggtccctcactctgtctggctggcactgatgaacaactatcaactcccttgatcgctg caattattgtgacccggaagctgcacccatccagcaattatttaattgttcccttgagtcaca gatttcttctgtgctgtcctggatgaccttcagcatgtgatatgtgagagagagctggat tatggggcaagtgtctgtgacatttggctgagtggtgacattacctgctgcagtgctccatct tgatctctcagctatagcttggatcggtatcgagcaatcacagatgctgtgagatgcccagg aaaaggactccaaagcatgctggcatttatgattacaatagttggatttatctgttttctc tatgctctctctatcttggaggcaccaaaggactagcagagatgatt/g]gaatgcacatcaag cagaccacattgttccacatttactcaacatttggagcttTGAGGCTTTTCTACATCCAC	640
HTR1Eu 1	HTR1E	T	C	I	T	cds	GE1157	CAGCCAAAGGA AAATAACCAA	GCACCCAGCGT GGAGTAAAT	CAGCCAAAGGAAATAACCAACagcttctccacagtgtagctgaacaagggaacatgaacat cacaactgtaccacagagggccagcatggctataagacccaagaccatcactgagaagatgctca tttgcatgactctgggtggtcatcaccacccctcaccagcttgctgactggctgtgatt/c]cat ggctattggcaccaccaagaagctccaccagctgcacactcaactatctgtctctggccgtga cggacctctgtggcagtgctgtctatgccccctgagcatcatcatatgtcatggatcgctgg aagcttgggtacttctctgtgaggtgtggctgagtgaggacatgacctgctgcacctgctccat cctccacctctgtgcatgccccctggacaggtactggccatcacaatgctattgaatacgcca ggaagaggacggccaaagggccgctgtagtacctaccgtctggaccatctccatcttctc tccatgccccctctgttctggagaagccaccgccccttaagccctccccctagtcagtgccaccat ccagcacgaccatgttactacacatttTACTTCCACGCTGGGTGC	630

FIG. 5Wwww

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
HTR1EU 2	HTR1E	G	T	E	D	cds	GE1156	GCACGACCATTGTTATCTACACC	CCCATCCACTC ATGAGGCT	GCACGACCATTGTTATCTACACCatttactccacgctgggtggcgttttatatcccccttgactttga tactgattctctattaccggatttaccacgcgcccaagagcctttaccagaaaaaggggatcaagt cggcacttaagcaacagaagcacagatagccagaattctttgcaagttgttaaaattacacagac ttctgtgtgtgacttctccacctcagacctaccacagagtttgaaagttccatgctctcca tcaggatcccccttcgacaattgatacagccagagacgttcagcagatctctcagcacc agggaaagggaagcagcagcatcctggggtgattctgggtgattcatttctcctggctgccc attttcatcaaga [g/t]ttgattgggtctgagcatctacacggtgctctcggaaagtggcc gactttctgacgtggctcggttatgtgaattctctgataacacctgctctatacagagttttaa tgaagactttaagctggcttttaaaagctcatatagatgccgagagcacttagactgtataaaa gttaaaaggcagcacttttccagAGCCTCATGATGGATGGG
HTR1EU 3	HTR1E	C	A	L	I	cds	GE1156	GCACGACCATTGTTATCTACACC	CCCATCCACTC ATGAGGCT	GCACGACCATTGTTATCTACACCatttactccacgctgggtggcgttttatatcccccttgactttga tactgattctctattaccggatttaccacgcgcccaagagcctttaccagaaaaaggggatcaagt cggcacttaagcaacagaagcacagatagccagaattctttgcaagttgttaaa [c/a]ttacac agactttctgtgtgtgacttctccacctcagacctaccacagagtttgaaagttccatgcc tccatcaggatcccccttcgacaattgatacagccagagacgttcagcagatctcttag caccaggaaacggaaaggcagcagcatcctggggtgattctgggtgattcatttctcctggc tgccattttcatcaaaagttgattgtgggtcagagcatctacacggtgctctcggaaagtggcc gactttctgacgtggctcggttatgtgaattctctgataacacctgctctatacagagttttaa tgaagactttaagctggcttttaaaagctcatatagatgccgagagcacttagactgtataaaa gttaaaaggcagcacttttccagAGCCTCATGATGGATGGG
HTR1EU 4	HTR1E	C	T	H	H	cds	GE1157	CAGCAAAAGGA AAATAACCAA	GCACCCAGCGT GGAGTAAT	CAGCAAAAGGAATAACCAACagcttctccacagtgtagctgaaacaaagggaacatgaacat cacaactgtaccacagagccagcatggctatagaacccagaccatcactgagaagatgctca tttgcatgactctgggtggtcatcaccacctcaccacgttgctgaacttggctggtatcatggct attggcaccacaaagaagctccaccagctcccaactacctaattcttctctggcgtgacgga cctcctgggtggcagtgctcgtcatgccccctgagcatcatcatattgtatggatcgctggaaagc ttgggtacttctctgtgaggtggctgagtgaggacatgacctgctgacctgctccatcctc cactctgtgcatgcccctggacaggtactgggccatcacaatgctatgaaacggccaggaa gaggacggccaaagggccgctgattgcttaccgttcggaccattccatttctcatctcca tgccccctctgtctggaaagcagccgcctaaagcctccccctccctagtcagtgaccatccag ca [c/t]gaccatgttatctacacatttTAC'TCCACGCTGGGTGGC
HTR1EU 5	HTR1E	C	T	S	F	cds	GE1156	GCACGACCATTGTTATCTACACC	CCCATCCACTC ATGAGGCT	GCACGACCATTGTTATCTACACCatttactccacgctgggtggcgttttatatcccccttgactttga tactgattctctattaccggatttaccacgcgcccaagagcctttaccagaaaaaggggatcaagt cggcacttaagcaacagaagcacagatagccagaattctttgcaagttgttaaaattacacagac ttctgtgtgtgtgacttctccacctcagacctaccacagagtttgaaagttccatgctc [c/ t]catcaggatcccccttcgacaattgatacagccagagacgttcagcagatctcttag caccaggaaacggaaaggcagcagcatcctggggtgattctgggtgattcatttctcctggc tgccattttcatcaaaagttgattgtgggtcagagcatctacacggtgctctcggaaagtggcc gactttctgacgtggctcggttatgtgaattctctgataacacctgctctatacagagttttaa tgaagactttaagctggcttttaaaagctcatatagatgccgagagcacttagactgtataaaa gttaaaaggcagcacttttccagAGCCTCATGATGGATGGG

FIG. 5XXXX

FIG. 5YYYY

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
HTR2Ad 14	HTR2A	C	T	-	-	noncoding	GE1167	TGGTACTGCGA AACCNA	CGCACTGCTAG GATCCTGTT	TGGTACTGCGAACCNAActtatttctaccacatgtgaggttttgaataataatctcgggtgycata ttctctgtgagagaaataaagccagttcaatgggtgtatctattataataataataatagtgtaaatagtt tatcagagttatcaccacagactgcctagccacctgagcctatgtggccaatgtcagtaaatctcc actctggacacaaacactgttggcttggatggagtgccagacactcacagcctc[c/t]gag gacatactgtttctagccttatttattgttctctcttccaccttcagcctaaaaattaccacag tgccacttacctaccttaattgggaatctgcacaaaggccatttctcagtttctctcaaaagca aaggaaaaacttcccaatattaatatgtagcaaaaggaggagaaagcctgtttgggtccgc cctcctggctgttgcctaccttgcagcttatgacatacacatagaggggtctgtgaaatgaac gagacagtcagagagctacctcccttccgtggaaacccagggctcccttgggtgcagacagctcttc ctacttcccatgcagtctcttctgtgcgacttttgggggctcgtgaatgttctaaatgtgtgc ctgctgaggcgagccgcacagggaggaggaaacccagccgagcgtgcccagaggagccAACAGG ATCCTAGCAGTGCG	729
HTR2Ad 15	HTR2A	G	A	-	-	noncoding	GE1169	CCGTGCCAGAG GAAGCC	CTCACCAACC GAGGACNA	CCGTGCCAGAGGAGCCaacaggatccttagcagtgcggaactggctcagctcttgcagtgcagttt ttgaagtcagcaaaacagaaaccaaatactactatcatattatgctggtggaagatcaagaagaggg gacttacaccagtttaattactgtgagagatgcagcgagtcacagaataacaaatgtatctcat gttgaaacctgaagacaaatgttagtctctcatgcgctatatttattgtgtgtaatttctctt tccgggttgaaatcatgcttgcccaaatgtaacttcaatgagaatttccaggggaggaaagt tgtctgttaatttacttaagactttttgtttctcttatttagctaagcaacttattagagag ctgaaattcctgacagcagctgtggcaattcagcctaagaaatggctgagaactgtaacccaaga tataccaattactatgggattaacactggatgtatttttaattgacttcttattgtagaatgt gtacatccccactgttctgattgcagctattttaataataactgttgcataactagtaccatcl g/a]gcataacccaacaaatgagatagtttaacaagagtcacagagttataaaactttctt cttctgccagaacatttattctccccgaacgctcaaaaaaaccttgcacctctatgctaaaaag tttcaattctgtcttTTTGTCCTCGTGTGTGAG	749
HTR2Au 1	HTR2A	G	C	-	-	noncoding	GE1167	TGGTACTGCGA AACCNA	CGCACTGCTAG GATCCTGTT	TGGTACTGCGAACCNAActtatttctaccacatgtgaggttttgaataataatctcgggtgycata ttctctgtgagagaaataaagccagttcaatgggtgtatctattataataataataatagtgtaaatagtt tatcagagttatcaccaca[c/g]actgcctagcaccctcgagcctatgtggccaatgtcagtaaa ttccactctggacacaaacactgttggcttggatggagtgccagacactcacagcactccgag gacatactgtttctagccttatttattgttctcttccaccttcagcctaaaaattaccacag tgccacttacctaccttaattgggaatctgcacaaaggccatttctcagtttctctcaaaagca aaggaaaaacttcccaatattaatatgtagcaaaaggaggagaaagcctgtttgggtccgc cctcctggctgttgcctaccttgcagcttatgacatacacatagaggggtctgtgaaatgaac gagacagtcagagagctacctcccttccgtggaaacccagggctcccttgggtgcagacagctcttc ctacttcccatgcagtctcttctgtgcgacttttgggggctcgtgaatgttctaaatgtgtgc ctgctgaggcgagccgcacagggaggaggaaacccagccgagcgtgcccagaggagccAACAGG ATCCTAGCAGTGCG	729

FIG. 5ZZZZ

FIG. 5A AAAA

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
HTR2AU 13	HTR2A	A	G	-	-	noncoding	GE1169	CCGTGCCAGAG GAAGCC	CTCACCACAAACC GAGGACAAA	CCGTGCCAGAGAGAGCCCaacaggatcctagcagtcgaggacgtggctcagctcttgcagctt 749 ttgaagtcagcaaaacagaaaccaaataactatactatgctggtggaagatcaagaagagggg gactctacaccagtttaattactgtgagagatgagcagcagtcacagatacaaatgtatctcat gtgtgaacccctgaagacaaaatgaagtctcatgccgtatatttattgtgtgtaatttctt tccgggtttgaaatcatgcttgcccaacatgtaacttccatttcattgagaaattccaggaggaaagt tgctgtcaatcttacttaagactttttgttttcttcttcttcttcttcttcttcttcttcttctt ctgaaattcctgacagcagctgaggcaattcagcctaaagatggctgaggaactgttaacccaaga tacctcaattactatgggattaacactggatgtatttttaattgacttctttaaagttagaattg gtacatccccacgtttctgattgcatgctattttaaataactgttgcataaactgttaccatcg gcataacccaacaaaatgagatagtttaaacacagagtcacagtagttataaaacttttcttctt gtccagaacatttatcttcccga a/g cgctcaaaaaaacccctgcaacctctatgctaaaag tttcaattctgcttTTTGTCCTCGGTGGTGAG
										TGGTACTGCGAACAACtaatttcttaccacacatgtgagggttttgaataataatctgggtggcata ttctgtcgaagaaataagccagttcaatgggtatctatactataataaaatagtgctaaatggt tatcagagttatcaccacagactgcttagccacccctgagcctatgtggccaatgtcagtaattcc actctggacacaaacactgttggttggatggaagtgc a/g gacactcacagcactccagag gacatactgtttctagccttatttattgtctctctctcctcagcctcaaaaattaccacag tgccacttacctaccttaattgggaatctgcacaaaaggccatttctccagtttctctcaaaagca aaggaaaacttcccaataatataatgtagcaaaaaggaggagaaaagcctgtttggtccgc cctcctggcgtgtgtctaccttgacgttatgacatacacatagaggaggtctgatgaatgaac gagacagtcagagagctactcctcctcgtggaacacagaggtcccttgggtgcagacagctcttc ctacttccccatgcagttcttctgtgcgactttgagggtgtcgtagaattcttaattgtgtgc ctgctgaggcgagccgcacaggaggagggaaccacagccgagcgtgccagagggaagccAACAGG ATCCTAGCAGTGCG
HTR2AU 2	HTR2A	A	G	-	-	noncoding	GE1167	TGGTACTGCGA AACCA	CGCACTGCTAG GATCTGTT	CCGTGCCAGAGAGCCCaacaggatcctagcagtcgaggacgtggctcagctcttgcagctt 749 ttgaagtcagcaaaacagaaaccaaataactatactatactataataaaatagtgctaaatggt gactctacaccagtttaattactgtgagagatgagcagcagtcacagatacaaatgtatctcat gtgtg a/g acctgaagacaaaatgaagtctcctatgcccctataatttattgtgtgtaattt tcttccgggtttgaaatcatgcttgcccaacatgtaacttcaattcgaatttccaggaggaga aagtgtctgtctatcttacttaagactttttgttttcttcttcttcttcttcttcttcttctt ggagctgaaattcctgacagcagctgaggcaattcagcctaaagatggctgagaaactgttaacca aagatacatccaattactatgggattaacactggatgtatttttaattgacttctttaaagttaga atgtgtacatccccactgtttctgtatgcatgctattttaaataactgttgcataaactgtacc atcggcataaaccaacaaaatgagatagtttaaacacagagtcacagtagttataaaacttttctt cttgcacagaaacatttatcttcccgaacgtctcaaaaaaacccctgcaacctctatgctaaaag tttcaattctgcttTTTGTCCTCGGTGGTGAG
HTR2AU 3	HTR2A	A	G	-	-	noncoding	GE1169	CCGTGCCAGAG GAAGCC	CTCACCACAAACC GAGGACAAA	CCGTGCCAGAGAGCCCaacaggatcctagcagtcgaggacgtggctcagctcttgcagctt 749 ttgaagtcagcaaaacagaaaccaaataactatactatactataataaaatagtgctaaatggt gactctacaccagtttaattactgtgagagatgagcagcagtcacagatacaaatgtatctcat gtgtg a/g acctgaagacaaaatgaagtctcctatgcccctataatttattgtgtgtaattt tcttccgggtttgaaatcatgcttgcccaacatgtaacttcaattcgaatttccaggaggaga aagtgtctgtctatcttacttaagactttttgttttcttcttcttcttcttcttcttcttctt ggagctgaaattcctgacagcagctgaggcaattcagcctaaagatggctgagaaactgttaacca aagatacatccaattactatgggattaacactggatgtatttttaattgacttctttaaagttaga atgtgtacatccccactgtttctgtatgcatgctattttaaataactgttgcataaactgtacc atcggcataaaccaacaaaatgagatagtttaaacacagagtcacagtagttataaaacttttctt cttgcacagaaacatttatcttcccgaacgtctcaaaaaaacccctgcaacctctatgctaaaag tttcaattctgcttTTTGTCCTCGGTGGTGAG

FIG. 5B BBBB

[illegible]

FIG. 5CCCC

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
HTR2Cu 2	HTR2C	A	C	M	L	cds	GE1275	CCTAGATATT GTGCCCGT	CGATCAACGC AAATGT	CCTAGATATTGTGCCCGTcttggtttctttagatgttttattttcaacagcgctccatc(a/c) 298 tgccctctgcgtatctcgctggatcggtatgagcaatagcaatcctatgagcatagccgt ttcaattcgaggactaagccatcatgaagatgctatgtttgggcaatttctataggtaata aaacttttggccataagaattgcagcggtatgctcaactttcggtattatgtactgtgaaca acgtacagacgtcgactggtaACATTGCTTTGATCG
HTR2Cu 3	HTR2C	C	G	L	V	cds	GE993	CAAAAGAAAG ATGATATGATG A	TTACAGGAATG AAATGCACCG	CAAAAGAAAGATGATATGAAactagcctgttaatttcgtctctcaattttaaactttggt 128 tgcttaagactgaagcaatcatggtgaac(c/g)tgaggaaatgCGGTGCATTCTCTGTAA
HTR2Cu 4	HTR2C	C	G	I	M	cds	GE1275	CCTAGATATT GTGCCCGT	CGATCAACGC AAATGT	CCTAGATATTGTGCCCGTcttggtttctttagatgttttattttcaacagcgctccatcagca 298 cctctgcgtatatacgctggatcggtatgagcaatagcaatcctatgagcatagccgtttca attcgcggaactaaggccat(c/g)atgaagatgctatgtttgggcaatttctataggtaata aaacttttggccataagaattgcagcggtatgctcaactttcggtattatgtactgtgaaca acgtacagacgtcgactggtaACATTGCTTTGATCG
HTR5Au 1	HTR5A	T	C	H	H	cds	GE1319	TCTGCAAGTAC CCCAGG	GGATTGCTGAG ATACCCA	TCTGCAAGTACCCAGGcggtctcctgaccagagatggatttacagtgaacctaaacctctt 800 ttccctctccacccctccctttggagaccacacacagcctcggaagacagacctgccccca gctcgccctgctctcggtcttcggatgcttatctacacttctggtggcggtcttctggtggcg acgttcgctggaaacctggtggtggggacacatcctcctgtagcagaccttccacggcggtcc ccacaacctggtggcatccatgctcgtctcgatgctcgttggtggcggtgctcagcgtgga gctggtgca(t/c)gagctgtcggcggtcgctggagctaggcgaggtgtgccaactttg gatcggtgagcgtgcttctgacgagccagcatctggaacgtgacggccatagccctggaacc gctactggtccatcacgcccacatggaaatacacgctcgcgaccccgagctgctccaaactc atgatcggtcactcactgggacactcctcgtctgctatctctggcccgctgcttttggctgggg agagacgtactctgagggcagcgagagtgccaggtgaagcggagccttctacgctgctct ccacgttaggccttctacactgcgctctgtgtgtggtctctgtagtactggaagatctacaag gctgccaagtctcggtgggtccaggagaacaaatagcgtctcaccatataccgaagctgtgga ggTGGGTATCTCAGCAATCC
HTR5Au 2	HTR5A	A	T	P	P	cds	GE1319	TCTGCAAGTAC CCCAGG	GGATTGCTGAG ATACCCA	TCTGCAAGTACCCAGGcggtctcctgaccagagatggatttacc(a/t)glgaacctaacct 800 cctttccctctccacccctccctttggagaccacacacagcctcggaagacagacctgggc cccagctcgccctgctctcggtcttcggatgcttatctacacttctggtggcggtcttctggtggc ggagacgttcgctggaaacctggtggtggtggggacacatcctcgttagcagaccttccacggcg tgccccacaacctggtggcatccatggcgtctcgatgctcctggtggcggtggtcactgccc ctgagcctggtgcatgagctgtcggggcggtggagctaggctggaggtgtgccaactttg gatcggtgagcgtgcttctgacgagccagcatctggaacgtgacggccatagccctggacc gctactggtccatcacgcccacatggaaatacacgctcgcgaccccgcaagtgcctccaaactc atgatcggtcactcactgggacactcctcgtctgctatctctggcccgctgcttttggctgggg agagacgtactctgagggcagcgagagtgccaggtgaagcggagccttctacgctgctct ccacgttaggccttctacactgcgctctgtgtgtgtggtctctgtagtactggaagatctacaag gctgccaagtctcggtgggtccaggagaacaaatagcgtctcaccatataccgaagctgtgga ggTGGGTATCTCAGCAATCC

FIG. 5EEEE

[illegible]

FIG. 5FFFF

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
HTR6u3	HTR6	C	A	P	T	cds	GE1148	CCAGGCCGCTGT GCGACT	AGGTCTGGGT TCTGCTCA	CCAGGCCGCTGTGCGACTGcattccccagggcctctcgatgtcctcacatggctgggttactgta acagcacatgaacccccatctaccacactcttcatgaggagccagcctggcctggcctggcaggg ttcctgccatgtccacgctgtccccggagcagccagcagcctgacagcggcgtggcaggg ctctcacagcggccccggcccccttagcctacagcaggtgctg[c/a]cgctggcctggcctg ccggactcagattcggactcagacgagcctcagcggcctctcggcctcggcctcagcggccca gtctgtcttctcggcagggcagccagggacccccggcctgcccacagggcctgcccggcgtca attcttcaacatcgacccccggagcagcagctgcccggcctccacattggcattccccacgaac tgacccgggcttggggctggccaaatgggagagctggatTGAGCAGAACCCAGACCCCT
HTR6u4	HTR6	C	T	A	A	cds	GE1148	CCAGGCCGCTGT GCGACT	AGGTCTGGGT TCTGCTCA	CCAGGCCGCTGTGCGACTGcattccccagggcctctcgatgtcctcacatggctgggttactgta acagcacatgaacccccatctaccacactcttcatgaggagcttcaagcgggctggcaggg ttcctgccatgtccacgctgtccccggagcggcagcagcctggcctcgccatcactgagcac ctctcacagcggccccggcccccttagcctacagcaggtgctgctggcctggcctcactgagcac actcagattcggactcagacgagcctcagcggcctctcggcctcggcctcagcggcctgagctg ctgtctctggcagggcagccagggacccccggcctgcccacagggcctgcccgc[c/t]gtca atttctcaacatcgacccccggagcagcagctgcccggcctccacattggcattccccacgaac tgacccgggcttggggctggccaaatgggagagctggatTGAGCAGAACCCAGACCCCT
HTR7d1	HTR7	T	C	L	P	cds	GE1178	TGCTTGCTTG ACCAGTTATG	AGTGATAAATG ACCTTACAGCA	TGCTTGCTTGACCGATTATGatcacgggtgttcactgtttacttccactctccactagggtac cttggggtcacagggccccctcacataccctgtgaggcagaaatgggaaatgcatggcgaagatgat ctctccgctctggtctctctccctccatcaccttaccctcactcttggatgggtcagaaatg taaatgatgataagggtgctgtgacagcagcaggttggctatacagatttactcaccgagtg gcattttatatccccatgtccgtcatgtttctgactacacagatttacaaggctgccagga gagtgctgcccacacacaaagtctctggcttccctggctgagtgaggagtgagcagcctcagga atggcatagtgaagctccagaggggtggaaggtgagcagccttcagagactctcagacat gaagggaacacatctccatcttaagcagagacagaaagcagccttcagagactctcagacat cggggcttaccgctgtgctggctggccttctccctcctcagcagcagccttcactctgtg gcactctctgagctgcatccactgtgggtggagagacattctgtggctaggctatgcaaac tctctcattaaccttttatatatgtcttctcaccgggacctgaggaccacacctatcgagcct gtccagtgccagtagccggaatatcaaccggagagctctcagctgcagctgagcagcctga agc[t/c]tgctgagaggccagagagacctgagttgtTGCTGAAGGTCAATTATGACT
IGF1d3 5	IGF1	A	G	-	-	noncoding	GE1192	GGCTTAATAAA ATAGCATTAGG T	GATGCCATTGC ATAATCAGA	GGCTTAATAAATAGCATTAGGTctattctagccaccacacacttcaactttttatcaccaaa gtagtactgttccacaaattgtgaatttgggggtgaggggaggggttggaatttttttaa gttagaagggtccattgtttgttggctctcaacttagcaaaatlagcaatatatatcca[ta/ gl]tctctgaacttgatcaagagcagtgagaaataacgcggggaaaaagatcttataggcaata gaagaatttaaaagataagtaagttccttattgtatttttgacactctgctctaaacagatatt cagcaagtggaagaaataagaacaaagagaaataacatagatttacctgcaaaaaatagcttc tgccaaantccccctgggaatccttggcaatttactggtttatagaagacattctccctcac ccagacatctcaagagcagtagctctcatgaaagcaatcactgactcatttgggaaatgttg gaagatttctctatgagatgggggttactactgataaagaaagaaatattatgagaaattgtg aaagagatggctaaccaattgtgaagattttttgttcttgggtttgtttttttttttttta ctttatcacagctcttatgaatttcttaattgttcaaaatgactgggttcttctcttttttta tatcagaatgaggaataataagtttaaacccacatagactcttaaaactataggctagatagaaa tgtatgtttgactgttgaagctataatcagactatttaaaatgttttgctatttttaattcttaa aagattgtgctaatattatagagcgaacccgtgttggtctctcctcagagaaagaaatcttccat tcaaatcacatgggtttccaccaataattttcaaaagataaataTCTGATTATGCAATGGCATC

FIG. 5GGGGG

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
IGF1d36	IGF1	C	T	C	C	cds	GE591	GAAAGCAGATTGCACCTAAC	CCACCCAGGTGGCTTA	GAAAGCAGATTGCACCTAACatgagccactctgttggattgtgtagacacagccacaggggtatggtctccagcagtcggagggcgccctcagacaggcatctgtgtagtgagtg c t tgctccggagctgtagatctaaggagctggagatgattgcgcacccctcagcctgccaagctcagctcgctctgtccgtgcccagcgccacaccgacatgcccaagacccagaggTAAGCCCACTGGTGG
IGF1d37	IGF1	C	T	-	-	noncoding	GE678	AGGGCGCTTGA GTTGCT	GCTGGATAATT CATTTGTTCTAA T	AGGGCGCTTGA GTTGCTgagatgcaagggaattctataataaacccattcatagcatagctagaga ttggtggaattgaaatgctcctcagcatctcagttcttctgtagtgagctatccaaataaactggccaa ctgagttgttaaaagctaaacagctcaatctcttaaaacacttttcaaaatattgtgggaagcatttg atttccaatttgatttgaattctgcaatttggtttttatgaatacaaaagataagtgaaagagagaga aaggaaaagaaaagagagaaaacaaagagatttctaccagtgaagggaattaatctactctt gttag c t actcactgactctctctatgcagttactacatatcttagtaaaccttgtttaatac tataataataattctattcttltgaaaacacacaaatgattcctcttcttaggcaataaagga aagtgatccaaaatttgaataattaaataataataataataaaagtcacaaagttaattctcttta aaaaacttactcttattcttagctgtatataacattttttaaagtttgttaaataatgcttg actagagtttcagttgaaaggcaaaaactccatcacaaaganaatttcccatgcctgctcaga agggtagccctagctctctggaatggttttctcattcactcaactgaaaattgggtatcaagaaag tccactgggttagtactgactcctcatagccttagaaaatgatccctatctgacagatcaagattt tctcATTAGAACAAATCAATTATCCAGC
IGF1ul	IGF1	C	A	A	D	cds	GE638	GGAACCACTTG TTCTCAATGC	TTTCCGTTTTC TCCATGTTTC	GGAACCACTTGTTCTCAATGCAattatttttggtaggtttacagttatcagcccccatctaccaac aagaacacgaagctcagagaaggaaaggttggccaaagacacatccaggagggggaacagaaagga ggggacagaagcaagctctgcagatcagagaagaagaagaagagagagagagagatttggaaagta gaaatg c/a tgaatgcagaggcacaagaaagggaatgaaggcagagagatttaaacagacagag gcaaggatgatgagagagagagagacagaagaatgaagaaagcagaaaatacaatagagagaaatga agaaagtaggcctgctggagctagatgatgtgatggaataagaagtaaccttttagagaat ctgcgtaaGAAACATGGAGAAACGGAA
IGF1ul0	IGF1	A	G	-	-	noncoding	GE688	TCATAGCCTAG AAAATGATCCC TAT	AGGGTTGTCAT CAATTGTTT	TCATAGCCTAGAAATGATCCCTATctgcagatcaagattttctcattagacaatgaattatcc agcattcagatcttcttagtcaccttagaacttttgggttaaaagtaaccaggttgattatctt atgcaaatctctatatttacctcttggaagctctatagaaacacaaataaactctcagtt tttctccactgggtcacctcaaggatcagaggccaggaacaaacaaaggaactccctggatctc tgaatatatgcaaaaagaaaggcccaatttagtgagccagcaaatctgtccagtcacaaagttt taaactcagtcacaacattatttgaattgagcacctcaagcatgcttagcaatgttttaatacac tatggacagatgtaaaagaaactatacatcttttggccctcgcctgtttccagacatacagg ttctgtggaataagatactggactcctctcccaagatggcactcttttatttcttctgtccca gttgtacctttttaaattattccctctcaacaaactttataggcagctcttctgcagacttaac a/g tgtttctcagatgtagatgataaattcaagatgctctatgacttatttctctcac ttaattctatccacagtcaaaatcccccaagggaagagtgaaagatgcaactgccaatatta tcttcttaacttttccaacacataaatctctccactggattataataaattgaaataaact cattataccaattcactattttattttttaaagaattaaactcagaAAACAAATTGATGCAAAACC CT

FIG. 5HHHHH

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
IGF1ul1	IGF1	T	G	-	-	noncoding	GE682	CCACACATAA TCCTCTCCAAC T	CACATTGGCAT AGCTGGC	CCACACATAATCTCTCCAACTGGattataaaataaattgaaaaataaactcattataccaattcac tatttatttttaattgaattaaaactagaaaaaaattgatgcaaaccttgaagtcagtcagtcgat (t/g)actatactacacagagaatgactcagatttcatagaagggaacacaaaatgtcacaa cgaactttacaagctttgtcttcagaattagattgctttataattcttgaaatgaggcaatttca agatatttgaaaagacagtaaacattggtaagaatgagctttcaactcataggcttatttcca atttaattgaccatactggatactttaggtcaaatctctgtctcttgcaccaataataataaa gtattattgacttttaagatgaggcagttcccttgaataaattgaatgcagctctccatcaga atccactcttctagggatatgaaaatctcttaacacccacctacatacacagacacacacac acacacacacacacacacacacacacattcccttaaggatccaatggaaatctgaaagaaaa tcaacttcttgaaaaattttattaaaaaaacaaacaaacaaacacacacacacacacacac tccctccctcccttggaagtcgaattgttctgttagatgaaacacacacacacacacacac ggttctgttcaactatttcatgcacttgggagaggcttagaataaaagatgtagcacatttgc tcccaatttattgttttggcagacttgcacacacacacacacacacacacacacacacacac	815
IGF1ul2	IGF1	T	C	-	-	noncoding	GE683	TTTTATATTAC TGAGCCCTAAA AGT	AGATATACCAT TTTATTATGAC ACTCT	TTTTATATTACTGAGCCCTAAAAGTaaacattactcatttatttggcccaaaatgcactgatgt aaagttagaaaaataaaaacagagctctaaaatcccttcaagccacacatttgacccactcacc aacctatagaaaagtcaactctctgttaactcccttaactgatctgtttggataattatctgtac ccgtctgctaaacacactgcaggaggacttgaacacctcaagctgtctacttactcttctatct gtgtctgtgtatcatgaaaatgtctattcaaaatatacaaaccttcaaatcaacgcagcttat attcagtttacataaaagggcccaataaccatgtcagatcttttggtaaaagagttaatgaacta tgagaattgggatcacatcatgtatttgcctcatgtattttatcaccttataagcagcagctg t/cjgataaaataaacttacagacactgaattaatctccctgtcactctgaaacagaaaaaat gactggccactcgttacactcgtcttagtgaagacataatttttataaaattaatctgattg tatttgaaattatttcaattcaactatggcagagggaatcaatcctaataatgacttcaaaaat gtaactaaattgaatcatatcttcaatttactgttttaataagcatttttgaaaaatgtatggcta GAGTGTCTCATATAATAAATGGTATATCT	741
IGF1ul3	IGF1	G	A	G	E	cds	GE642	AAAATGCTTC TGTGCTCTAGT	GAATTCGCCAA TGACTTCAA	AAAATGCTTCCTGTGCTCTAGTttttaaataatgcaaaaggatgatgttatttgtcaccatgcccaaa aaagtccttactcaataactttgcagaaaggaggagaggaaggcaaaatgttccccagct gtttctctgtctacagtcgtgtgtttgtagataaattggaggatcttctctaaatccccctct gtttgctaaatctcaactcactgcactgaatcaatcgagcagantagagcctgcgaatgaaataag tccctcaaaattgaaatgtgacatgtcttcaacatctccatctctctggatttttttctcttc attattccctgtcaacaaattcatttccagacttgcacttcagaagcaatgg(g/a)aaaaatca gcagctcttccaaaccaaattatttaagtgctgtcttcttgatcttcttgaaaggttaaatattctac tctTTGAAGTCATTGGGAATTC	478

FIG. 511111

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
IGF1u1 ₄	IGF1	T	G	-	-	noncoding	GE688	TCATAGCCTAG AAAATGATCCC TAT	AGGCTTTGTCAT CAATTGTGTTT	TCATAGCCTAGAAATGATCCCTATCTGcagatcaagattttctcattagacaatgaattatcc agcattcagatctttcttagtcaccttagaactttttgggttaaaagtagccaggcttgattatttc atgcaaa [t/g] tctatatcttacctcttggaagctctatagaaaaaataaacatcttc agttttctccactgggtcacctcaagatcagagccaggagaaaaaaagactccctgga tctctgaatatatgcaaaaagaggcccatctagtgagccagcaatctctcagtcacaacag tattttaactctcagtcacaacatatttgattgagaccccaagcatgcttagcaatgttctaa tcaactatgacagatgtaaaagaaactatacatcttttgcctctgcctgtttccagacata cagggtctgtggaataagatactggactctctctcccaagatggcactcttttcttctgtc cccagtgtagctaccttttaaaatttctctctcaaaaactttataggcagctctctgcagact taacatgtttctgtcatagtgatgtgataaattctcaagagtgctctatgacttattctctcac ttaattctatccacagtcacaaatcccccaaggaggaaagctgaagatgcaactgccaatatta tctttcttaacttttcccaacacataatctctccactggattataaataatgaaataaact cattataccaattcactattttattttttaaagaattaaaactagaAAACAAATTGATGCAAAACC CT
IGF1u1 ₅	IGF1	T	G	-	-	noncoding	GE688	TCATAGCCTAG AAAATGATCCC TAT	AGGCTTTGTCAT CAATTGTGTTT	TCATAGCCTAGAAATGATCCCTATCTGcagatcaagattttctcattagacaatgaattatcc agcattcagatctttcttagtcaccttagaactttttgggttaaaagtagccaggcttgattatttc atgcaaaattctatatcttacctcttggaagctctatagaaaaaataaacatctcagtt tttctccactgggtcacctcaagatcagagccaggagaaaaaaagactccctggatctc tgaatatatgcaaaaagaggcccatctagtgagccagcaatctctctcagtcacaacagattt ttaactcagtcacaacatatttgattgagcaccacagca [t/g] gcttagcaatgttctaa tcaactatgacagatgtaaaagaaactatacatcttttgcctctgcctgttttccagacata cagggtctgtggaataagatactggactctctctcccaagatggcactcttttcttctgtc cccagtgtagctaccttttaaaatttctctctcaaaaactttataggcagctctctgcagact taacatgtttctgtcatagtgatgtgataaattctcaagagtgctctatgacttattctctcac ttaattctatccacagtcacaaatcccccaaggaggaaagctgaagatgcaactgccaatatta tctttcttaacttttcccaacacataatctctccactggattataaataatgaaataaact cattataccaattcactattttattttttaaagaattaaaactagaAAACAAATTGATGCAAAACC CT
IGF1u1 ₆	IGF1	T	C	-	-	noncoding	GE688	TCATAGCCTAG AAAATGATCCC TAT	AGGCTTTGTCAT CAATTGTGTTT	TCATAGCCTAGAAATGATCCCTATCTGcagatcaagattttctcattagacaatgaattatcc agcattcagatctttcttagtcaccttagaactttttgggttaaaagtagccaggcttgattatttc atgcaaaattctatatcttacctcttggaagctctatagaaaaaataaacatctcagtt tttctccactgggtcacctcaagatcagagccaggagaaaaaaagactccctggatctc tgaatatatgcaaaaagaggcccatctagtgagccagcaatctctgtcagtcacaacagattt ttaactcagtcacaacatatttgattgagcaccacagcaatctctgtcagtcacaacagattt tatggacagatgtaaaagaaactatacatcttttgcctctgcctgttttccagacatacagg ttctgtggaataagatactggactctctctcccaagatggcactcttttcttctgtcccc g [t/c] gtgtaccttttaaaatttctctctcaaaaactttataggcagctctctgcagact taacatgtttctgtcatagtgatgtgataaattctcaagagtgctctatgacttattctctcac ttaattctatccacagtcacaaatcccccaaggaggaaagctgaagatgcaactgccaatatta tctttcttaacttttcccaacacataatctctccactggattataaataatgaaataaact cattataccaattcactattttattttttaaagaattaaaactagaAAACAAATTGATGCAAAACC CT

FIG. 5JJJJJ

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
IGF1ul	IGF1	A	T	-	-	noncoding	GE1192	GGCTTAATAA ATAGCATTAG T	GATGCCATTGC ATAAATCAGA	GGCTTAATAATAGCATTAGGTCATCTAGCACCACCACCTTTCAACTTTTATCACTCACAA gtagtgactgttcaccaaatgtgaatttcggggtgcaggggcaggagttggaaattttttaa gttagaaggctccattgtttgttggtctcaaaacttagcaaatatagcaatatattatccaatc ttctgaaacttgatcaagagcatggagaataaacgcggaataaagatcttataggcaaatggaag aatttaaagataagtaagttcccttatgtttttgtgactctgctctaaaacagatatccagc aagtggaagaaataagaacaaagagaaaaatacatagattacctgcaaaaatagctcttcgcc aaantccccntggggaatccttggaatttaactgggtttatagaagacattcctccctcacccag acattcgaagagcagtagctctcatgaaagcaatcacgtactctatttgggaattgttgaaa gtattccttatgagatgggggttatctactgataaagaagaatttatgagaattgttgaaag agatggctacaatctggaagatttltgtttcttggtttgttttttttttttttttttttt [a/e] tacagcttttatgaattcttaagtccaatagacttgggtctttctcttttttttt tatcagaatagggaataataaagttaaacccacacatagactctttaaaactataggatagaaa tgtatgttgacttgtgaagctaatatcagactatttcaaatgttttgcatttttaacttaa aagatttgcataatttatagagcagaacctgtttggctctctccacagaagaagaattcttccat tcaaatcacatggctttccaccaattttcagaagataaactgATTATTCATGGCATC	972
IGF1ul	IGF1	G	A	-	-	noncoding	GE683	TTTATATTAC TGAGGCTTAA AGT	AGATATACCAT TTTATTATGAC ACTCT	TTTATATTACTAGGCTTAAAGTaaacattactcatltttttggccaaaatgcactgatgt aaagttagaataataaaacagagctctaaaatcccccttaagccacacattgacccactcac aaactcatagcaaaagtcaactctgttaactcccttaactgtatttgtttgggatatattatctgtac ccgtgctaaacacactgcaggaggagctctgaaacctca[g/a]ctgtctacttccactcttt atctgtgtgtgtatcatgaaaatgctctatcaaaaatacaaaaaccttcaaatatcacgcagc ttatatctagtttacataaaggcccaaataccatgtcagatcttttttgggtaaaagattaatga actatgaaattgggattacatcatgtatttgcctctatgtttttatcacactatagggccaa gtgtgataaaataaaacttacagacactgaaataatattccccctgctacttgaacccagaaaataat gactggccattcgtttacatctgtcttagttgaaagcataatttttaataatttaattctgattg tatttgaaatttatattcaattcacttatggcaggaggaatatcaatcctcaatgacttctaaaaat gtaaatattgaatttatcttcaatttactgtttaaataagcatatttggaaaatgtatggcta GAGTGTCAATAAATGGTATATCT	741
IGF1ul	IGF1	C	G	-	-	noncoding	GE1191	TTTATAGGAAG TACATTGGAAG AAC	ACAAACTCAA AATAGCACCAT	TTTATAGGAAGTACATTGGAAGAACcaagttagagggagtgagggaacaaagaactcacaggatgt aggaaacccctctgagggggtgaaggtgacatgccaccgcaggatcctttgtctgcaacaggtt acctgttaaaactttggaacacacctaccaaaaataaagtttgataacaatttaaaagatggcggttc ccccaatgaaatacacaaagtaaaacatcccaaatgtcttaggaaggtgatttgcaccttgcaaaa atggtcctggagttggtagattgctgttgatctttatcaaatatgtcttatagaagaagaaaaa aaatataataataataatactttagtccctgctctcaagagccacaataatgcattgggtgtctgt atagatccagttgcactaaatt(c/g)ctctctgaattcttggctgtggagccattcattccagca accttgtctaagtggtttatgaattgttctctatttgcaactcttctcaacaactcgggctgt ttgtttacagtgctgataaatttgttagtcttatcccaacctccttcaataacctttatat ttggcgaatttggcctcctcaaaagcagcagcagtcgtcagaagcacacccaattcttaacccac aagattccactctgtggcatttgtaccaataatgaattggatgcattttttttagacacaaagct ttaatttccacatcatgcttacaanaaagaataatgcaaatagttgcaacttttgaggccaatca tttttaggcataatgttttaacatagaagtttcttcaactcaaaagagttccttcaaatgatga gttaaatgtgcaacctaaattagtaaccttccctcttttttttttcccatatagagcactatgtaa tttagcatatcaattatcacaggatatatcaaacagtatgtaaaacctgttttttttagtataATGG TGCTATTTTGTAGTTGT	993

FIG. 5KKKK

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
IGF1u2 ₂	IGF1	T	A	-	-	noncoding	GE1191	TTTATAGGAAG TACATTGGAAG AAC	ACAAACTACAA AATAGCACCAT	TTTATAGGAAGTACATTGGAAGAACGcaagtagagggtgcaggaaacaaagaactacagagatgt aggaagacccctctgaggagtgagagtgacatgccaccgcaggatcccttgcctcgcacgagtt acctgttaacctttggaacacctaccacaaaataaagtcttgataaacatttaaagatgggcgtttc ccccaatgaaatacacaaagtaaacattccaacattgctcttaggagtgatttgcaccttgcaaaa atggctcctggagttggtagattgctgtgatctttatacaaatgattcttatgaaaaagaaaaa aaatatataatataatataatcttagtccctgcctcaagagccacaaaatggatgggtgtgtgt atagatccagttgcataaatctctctggaatcttggctgggagccatctcatcagaaacct tgtctaagtggtttatgaattgtttccttattctgcaactctttctacacaaactggctgttgt ttcacagtgctgataaactgttagtctatacccaacctccctcataaccttatatttgc cgaattggcctctcaaaagcagcagcaagtcgtcaagagccacacaaatctcaacccacaaga ttccatctgtggcatttgtaccaaa[t/a]ataagttgggtgcatcttattttagacacaaaagt ttattttccacatcatcttcaaaaaaagaataatgcaaaatagttgcaactttgagggccaatca tttttaggcataatgttttaaacatagaaagtctcttcaactcaaaagagttccttcaaatgatga gttaattgtcaacctaaattagtaacttccctcttttatttttccatatagagcactatgtaaa tttagcatatacaattatacaggatataatcaaacagtatgtaaaaactgttttttttagtataATGG TGCTATTTTGTAGTTTGT	993
IGF1u2 ₃	IGF1	T	C	-	-	noncoding	GE1191	TTTATAGGAAG TACATTGGAAG AAC	ACAAACTACAA AATAGCACCAT	TTTATAGGAAGTACATTGGAAGAACGcaagtagagggtgcaggaaacaaagaactacagagatgt aggaagacccctctgaggagtgagagtgacatgccaccgcaggatcccttgcctcgcacgagtt acctgttaacctttggaacacctaccacaaaataaagtcttgataaacatttaaagatgggcgtttc ccccaatgaaatacacaaagtaaacattccaacattgctcttaggagtgatttgcaccttgcaaaa atggctcctggagttggtagattgctgtgatcttttatacaaatgttctatatgaaaaagaaaaa aaatatataatataatataatcttagtccctgcctcgaagagccacaaaatgcattgggtgtgtgt atagatccagttgcactaaatctctctgaaatcttggctgctggagccattctcagcaacct tgtctaagtggtttatgaattgtttccttatttgcactctcttctacacaaactcgggtgttgt ttcacagtgctgataaactgttagtctatacccaacctccctcataaccttatatttgc cgaattggcctctcaaaagcagcagcaagtcgtcaagagccacaccaaattcaacccacaaga ttccatctgtggcatcttgaccaaaataaagtgtgagtgcatcttattttagacacaaagctttat tttccacatcatgcttcaaaaaaagaataatgcaaatagttgcaactttgagggccaatcatctt taggcataatgttttaaacatagaaagtcttcaactcaaaagagttcttcaaatga[t/c]ga gttaattgtcaacctaaattagtaacttccctcttttatttttccatatagagcactatgtaaa tttagcatatacaattatacaggatataatcaaacagtatgtaaaaactgttttttttagtataATGG TGCTATTTTGTAGTTTGT	993
IGF1u2 ₄	IGF1	T	G	-	-	noncoding	GE683	TTTTATTATTAC TGAGGCCTAAA AGT	AGATATACAT TTTATTATGAC ACTCT	TTTTATTATTACTGAGGCCTAAAAGTaaacattactcatcttttattttgcccacaaaatgcactgatgt aaagtaggaaaaataaaaaacagagctctaaaaatcccttcaagccacccattgaccccaactcacc aaactcatagcaaaagtcactcttgttaactcccttaactctgatttggtttgatatttattcttgtag ccgctgtctaacaacacactgcaggaggagctctgaacacctcaagctgtctactacatcttttact gtgtctgtgtatacatgaaaaatgtctatacaaaatacaaaacctttcaaatatcacgcagcttat attcagtttacataaaggccccaaataccatgcatcttcttggtaaaagagtttaataaacta tgagaattgggaattacatcatgtattttgcctcatgtatttttatcacacttatagggccaagtgt gataaaataaacttacagacactgaaataatttccctgctacttttgaaaccagaaaaaatgaatgact ggccattcgtttacatctgtcttagttgaaaagcatttttttaataataattctgtattgtatt tgaaattattattcaattcacttatggcagagggaataatcaacttaaatgacttcttaaaaatgtaa ctaaattgaatcatta[t/g]cttcaacttactgttttaataagcatattttgaaaaatgtatggcta GAGTGTGCATAAATGGTATATCT	741

FIG. 5MMMM

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
IGF1u2 5	IGF1	T	C	-	-	noncoding	GE1192	GGCTTAATATA ATAGCATTAGG T	GATGCCATTGC ATAAATCAGA	GGCTTAATAAATAGCATTAGGTTCTATCTAGCCACCACCACCTTTCACCTTTTATCCTCACA gtagtgtactgttccacaaattgtgaatttgggggtgcagggtgagagtggtgaaattttttaa gtcagaaggctcccatgttttggctctcaacttagcaaaatagcaaatatattatccaatc ttctgaacttgatcagagcatggagatcaacgcgggaaagatcttataggcaaatagaag aatltaaaagataagtaogttccctattgttttggcactctgcttaaacagatatccagc aagtgagagaaataaagacaagagaaaaatacatagatttaccggcaaaaatagcttctgccc aaantccccctgggaatccttgcaatttactggtttatagaagacattctccctcaccag acattccaagagcagtagctctcatgaaaagcaatcactgactcttgggaaatgttggaaa gtatttcccttatgagatggggttatctactataaagaaagaaatttatgagaaatgttgaag agatggctaaacaatctgtgaagatttttggttcttgggtttgtttttttttttttttttt atacagctcttatgaatttcttaagtccaatgacttgggtcttcttcttcttcttcttctt agaatgggaataataagttaaacccacacacacacacacacacacacacacacacacac tgatgtttgactgttgaagctataatcagactctttaaagatttggctatttttaatttaa aagattgtgctaatltaagagcagacacacacacacacacacacacacacacacacacac tcaaatcacatgggttccac AGATAAGACAGAGCCAGGGgatttttgaagctgtcttattctgcccccatcccaaccagcc cttattatttttagtactgctcagaaattttatagagggtgacacagctgaaactctagaatta aaggaaacctcactgaaacacataatttcacgtgttccctctcttcttcttcttcttctgagatg gggtctcgcactgtccccgggtggagtgagtgagtgagtgagtgagtgagtgagtgagtgag tccctgggttgaagcagatctcctgctcagcctcctgagtgagtgagtgagtgagtgagtgag tatgccccgctaatttttggatttttaataagagacgggttttaccatgttggccaggttggag tcaaatctcctgacctgtgttggctcctcctcagcctcctcctcctcctcctcctcctcctc ccac ccttctcctcctcctcctcctcctcctcctcctcctcctcctcctcctcctcctcctcctc tttaagtgagagacagagtttctcctcaataacccgagctgaatttacccttcaaaaacacatg accttccac ATTTTATTGCCCCA	972
IGF1u2 6	IGF1	G	A	-	-	noncoding	GE676	AGATAAGACAG AGGCCCAGG	TTGGGCAAAAT AAATGAGTAA TGT	AGATAAGACAGAGCCAGGGgatttttgaagctgtcttattctgcccccatcccaaccagcc cttattatttttagtactgctcagaaattttatagagggtgacacagctgaaactctagaatta aaggaaacctcactgaaacacataatttcacgtgttccctctcttcttcttcttcttctgagatg gggtctcgcactgtccccgggtggagtgagtgagtgagtgagtgagtgagtgagtgagtgag tccctgggttgaagcagatctcctgctcagcctcctgagtgagtgagtgagtgagtgagtgag tatgccccgctaatttttggatttttaataagagacgggttttaccatgttggccaggttggag tcaaatctcctgacctgtgttggctcctcctcagcctcctcctcctcctcctcctcctcctc ccac ccttctcctcctcctcctcctcctcctcctcctcctcctcctcctcctcctcctcctcctc tttaagtgagagacagagtttctcctcaataacccgagctgaatttacccttcaaaaacacatg accttccac ATTTTATTGCCCCA	796
IGF1u2 7	IGF1	T	G	-	-	noncoding	GE682	CCAACACATAA TCCTCTCCAAC T	CACATTGGCAT AGCTGGC	CCAACACATAACTCTCTCCAACCTgattataaaataaattgaaataaactcatataccaattcac tattttattttttaagaaattaaaactagaaacaaatgtgatgcaaaccttggaagtcagttgat tactatatactacagcagaatgactcagatttcatagaaggagcacaacaaatgtcacacacaa aactttacaagcttgcctcagaattagattgctttataattcttgaatgaggcaatttcaagat atttgtaaaagaaacagtaaacatcgttgaagatgagcttccaactcaggtctatttccaattt aatgaccatactggatacttgggtcacaatttctgtctctctctctctctctctctctctctct ta(t/g)ttgaactttttaagatgaggcagttccccctgaaaaagtttaatgcagctctccatcaga atccactctctagggataatgaaatctcttaacacacacacacacacacacacacacacacac ac tcaacttccctgaaaatttttataaaaaacacacacacacacacacacacacacacacacacac tcccttccctccttggaaactgtttgtgtagatgaaacacacacacacacacacacacacacac gggttctgttactattttatgcacttgggagaggttagaataaagatgtagcacatttttgcct ttcccatatttattgttggccagctgagctgagctgagctgagctgagctgagctgagctgagct	815

FIG. 5NNNNN

[illegible]

FIG. 5PPPPP

[illegible]

FIG. 5QQQQ

[illegible]

FIG. 5RRRRR

[illegible]

FIG. 5SSSS

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
IGF2u1	IGF2	A	G	-	-	noncoding	GE689	GGAGAGCGTG GAGAGCA	CCAAATTGTTTC TCAGCCCAAT	GGAGAGCGTGAGAGCGAGGCACTGCTGACACGCGCCCTCCCTCCAGGACAACCTCCCCAGA TACCCGTGGGCAAGTCTCTCAATATGACACCTGGAAGCAGTCCACCCAGCGCTCGCAAGGG CCTGCTGCTCTCTGTCGCGCGGGGTCAGTGTCTGCGCAAGAGCTCGAGGCGTTCAGGG AGGCCAAACGTCAACGTCCTCTGATGCTCTACCCACCAAGACCCCGCCAGGGGGCGCCCC CCAGAGATGGCCAGCAATCGGAAGTGAAGCAAACTGCGCAAGTCTGAGCCCGCGCCACCATC CTGAGCTCTCTGACACGAGAGTTCATCAGGTTCATCCCGAAGAACTCTCTCGGTTCCAC GTCCCTGGGGTCTCTGACCCAGTCCCGTGGCGGCTCCCGAAGAGGCTACTCTCTCTC GGCCCTCCCTC/GTCTGGGCTGAGGAGACAGCAGCATCTTCAACATGTACAAATCGATTG GCTTAAACACCTTCACATACCTCCCTCCCAATATCCCAATATCCCAACATATAAAAT CAAAACATTAACTAACTCCCTCCCTCCCAACCAACCTCTTAAACATTAATGGCTTTT TAGAAACACCCCAAGCTCAGAAATGGCTTTAAAGAAACCAACCAACCAAAATCAAT GGCTAAAGAAAGTATTAAAGCAATTGGCTGAGAAACAATTGG	763
KLK2d1	KLK2	T	A	-	-	noncoding	GE478	GGCCAAAGAA CCAGGTG	CCAGAAATTAGG GTTTCACTCA	GGCCAAAGAAACCCAGGTGGGTCGGCCACAGCCAGT/a]tttctctgacccatagtcttgc gcccaggagttctcagtggtgagctccatctctctgccaatgacatggtgctagagcttac tctgagaaggtagacagagttcatgtgtgtgctgggctctggacaggtggttaagacacacttgg ggtagagtcactcctactcctcaacatctggagggggaaaggTGAGTGAACACCTTAATTCGG	256
KLK2d2	KLK2	A	G	-	-	noncoding	GE478	GGCCAAAGAA CCAGGTG	CCAGAAATTAGG GTTTCACTCA	GGCCAAAGAAACCCAGGTGGGTCGGCCACAGCCAGT/a]gttctctgaccc[a/g]tagtcttgc gcccaggagttctcagtggtgagctccatctctctgccaatgacatggtgctagagcttac tctgagaaggtagacagagttcatgtgtgtgctgggctctggacaggtggttaagacacacttgg ggtagagtcactcctactcctcaacatctggagggggaaaggTGAGTGAACACCTTAATTCGG	256
KLK2d3	KLK2	G	C	V	L	cds	GE484	CCCTCCCTAT CCAAATTCTTT	GCTGTTATTCC TGTTGGGACAC	CCCTCCCTATCCCTATCTTTTGGGTCGTCTGATCCCTGACCCAGCCCTCCCGAGGTGCTC /c]tgccctctcagctctcggattgtggaggctgggaggtgtagaagcattccccacccctgg cagtggtgctgtacagtcagtggtgacacactgtggggtgctctggtgacccccagtggtg gtctcagctgccccattgcttaagaagtaagttaggagccctgggattctggggagggaatggctgt GTCCACACAGGAATAACAGC	279
LIPCd2 5	LIPC	C	T	-	-	noncoding	GE253	CACACTGGACC GCAAAAGG	AGTGTGTGAGT TATTAGGCATG G	CACACTGGACCGCAAAAGGCTTTCATCCAGGCGAGTCTCTCTGCCCCCATCC/c/t]gctgc tgcttccagggaatctgttcaactctctcgaagccatgttccctaatgggtacagcctgggtg cacagtgctcaggatttgcgggcagttccatcgtggaaacgacacagattgggaggaatcacaggt aaCCATGCCCTAATAACTCACACCT	220
LIPCd2 6	LIPC	C	A	T	T	cds	GE335	CACCTCAATAA GCTCCACCTAA A	TCAGT-TTATGA GAGTGAACAT ACCC	CACCTCAATAAGCTCCACCTAAAACTTAAGGCTGTGTTGCTCTCTGTTTCTATTCCAGAAAGAC ATTTGTTCAAGAAACACAGATGACCTTACTCTGCCCCAAGGAGGAAATCTCTGTTGAAAT AATGTGAATAAAGTCTAAACATCAAGCGAAGATCAGATGAGATTAAATGAAGACCCAGT TAAAGATAAATGAATCTTCTCTTATCTGGAATGGTGTGCTTATTAGAGCCAAATACAT AAAGAACTCTCACAAAGCTTAAATAAAGTTAGATTAAAGGGGGTATGTTTTCACCTCATAAA CTGA	329
LIPCd2 7	LIPC	C	T	-	-	noncoding	GE335	CACCTCAATAA GCTCCACCTAA A	TCAGT-TTATGA GAGTGAACAT ACCC	CACCTCAATAAGCTCCACCTAAAACTTAAGGCTGTGTTGCTCTCTGTTTCTATTCCAGAAAGAC ATTTGTTCAAGAAACACAGATGACCTTACTCTGCCCCAAGGAGGAAATCTCTGTTGAAAT GTGAATAAAGTCTAAACATCAAGCGAAGATCAGATGAGATTAAATGAAGACCCAGTGTAAA GAATAAATGAATCTTCTCTTATCTGGAATGGTGTGCTTATTAGAGCCAAATACAT AAAGAACTCTCACAAAGCTTAAATAAAGTTAGATTAAAGGGGGTATGTTTTCACCTCATAAA CTGA	329

FIG. 5TTTTT

FIG. 5UUUU

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
LIPC ₂	LIPC	T	A	M	K	cds	GE353	AGCAGCCTTTG AGAAGACG	TCACAGCTCC CTATCTTTT T	AGCAGCCTTTGAGAGACGGgggttcagatgaagcagatgcaggctaacgacggtccccaat cttatatgcagagccatttgaagaagagctcaagctgttgaacaaacaaacgctgcagatgag alt/a)gaagaccagattctctcttggagaaacaaacagggctgtcagattcgaatcaatc atccggacacgttacaggagtgaggcttcaactctccctctggtgatgataatccacggg tggtcggtaggaaatgctgacatgccgttttctctccgatttcactagcgtgttcattcATA AAAAGATAGGGAGCTGGTGA
LIPC ₃	LIPC	G	C	G	A	cds	GE253	CACACTGGACC GCCAAAGG	AGTGTGTGAGT TATTAGGCATG G	CACACTGGACCGCAAAAGGcttctcatccaggcagctcttctctgccccccatcccgtgctgtc ttccaggaaatctgttcaactctctcgaagccatgttcaactaatgggtacagcctgggtgcaca cgtgtcaggatttgcg[g/c]cagttccatcgggtgaacgcacaagattgggagaatcacaggt aaCCATGCTTAATACTCACACACT
LIPC ₄	LIPC	A	T	Q	L	cds	GE475	AACTGATTGTG TCTGATTCTTCT	TGGCACAAGTG GGTGCTTA	AACTGATTGTGCTGATTCTTcttctgaaggggcaaggaattgctagtataaataaacgta ttctctttctatcagctggatggatctcggcgaggtgatcatgatcaagttcaagtgaggaaa acagtgagtggtggccaaatgtctgggacacgggtccagaccatcatccatggagcacagggccg cgccactcaggtctgttggaacgcatcagagtcgaagcgaagaaacccagc[a/t]aaggt gactgctgattcaatctctcttatacgtccatTAAGCACCCACTTGTGCCA
LIPC ₃	LIPC	T	G	V	V	cds	GE253	CACACTGGACC GCCAAAGG	AGTGTGTGAGT TATTAGGCATG G	CACACTGGACCGCAAAAGGcttctcatccaggcagctcttctctgccccccatcccgtgctgtc ttccaggaaatctgt[t/g]caactctctcgaagccatgttcaactaatgggtacagcctgggtg cacacgtgtcaggatttgcggcaggttccatcgttggaacgcacaagattgggagaatcacaggt aaCCATGCTTAATACTCACACACT
LIPC ₄	LIPC	T	A	L	Q	cds	GE253	CACACTGGACC GCCAAAGG	AGTGTGTGAGT TATTAGGCATG G	CACACTGGACCGCAAAAGGcttctcatccaggcagctcttctctgccccccatcccgtgctgtc ttccaggaaatctgttcaactctctcgaagccatgttcaactaatgggtacagcct[t/a]gggtg cacacgtgtcaggatttgcggcaggttccatcgttggaacgcacaagattgggagaatcacaggt aaCCATGCTTAATACTCACACACT
LIPC ₅	LIPC	A	G	G	G	cds	GE323	CTCCCGCGTAA CCCTTACC	CGGCCCATGAC TTCATTCTC	CTCCCGCGTAACCTTACCctctcttccatttgggtggtgagtcgagcggg[a/g]ccttggttt gagggagtgccccagcaaatctcttctccagatgatgcca[a/g]tttctggtatgccattcatatc ctttaccgggagacacatggcctgagcgtgggcatcaaacagcccataggacactatgacttct atcccaacgggggctcttccagcctggtgccacttcttagagctctacagacatatggcccag cacggcttcaatggTGAGAAATGAAGTCATGGGCCG
LIPC ₆	LIPC	A	G	N	S	cds	GE323	CTCCCGCGTAA CCCTTACC	CGGCCCATGAC TTCATTCTC	CTCCCGCGTAACCTTACCctctcttccatttgggtggtgagtcgagcgggaccttggttgagg gaagtgcctccagcaaatctgttcttccagatgatgcaa[t/g]tttctggtatgccattcatatc ctttaccgggagacacatggcctgagcgtgggcatcaaacagcccataggacactatgacttct atcccaacgggggctcttccagcctggtgccacttcttagagctctacagacatatggcccag cacggcttcaatggTGAGAAATGAAGTCATGGGCCG
LIPC ₇	LIPC	T	G	N	K	cds	GE323	CTCCCGCGTAA CCCTTACC	CGGCCCATGAC TTCATTCTC	CTCCCGCGTAACCTTACCctctcttccatttgggtggtgagtcgagcgggaccttggttgagg gaagtgcctccagcaaatctgttcttccagatgatgcaa[t/g]tttctggtatgccattcatatc ctttaccgggagacacatggcctgagcgtgggcatcaaacagcccataggacactatgacttct atcccaacgggggctcttccagcctggtgccacttcttagagctctacagacatatggcccag cacggcttcaatggTGAGAAATGAAGTCATGGGCCG
LIPC ₈	LIPC	C	G	T	T	cds	GE323	CTCCCGCGTAA CCCTTACC	CGGCCCATGAC TTCATTCTC	CTCCCGCGTAACCTTACCctctcttccatttgggtggtgagtcgagcgggaccttggttgagg gaagtgcctccagcaaatctgttcttccagatgatgcaaatttctggtatgccattcatatcctt ac[c/g]cgggagacatggcctgagcgtgggcatcaaacagcccataggacactatgacttct atcccaacgggggctcttccagcctggtgccacttcttagagctctacagacatatggcccag cacggcttcaatggTGAGAAATGAAGTCATGGGCCG

FIG. 5WVVVVVV

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
MPLu1	MPL	C	T	A	V	cds	GE929	CTGGTCCCTTC CCCTTC	CTGGAGTGGGA CTCAGC	CTGGTCCCTTCCTCCCTTCacataaactgcctggagagaccagggcccaactcaccagctgtccc ttagatgtctctctgtggtggtacacagactcagagccctgaaggttctcccgaacatttgagg cctcactgtctctgggatgaggagagggcag[c/t]gcccagtgggacataaccagctgtgtat gcctaccgcggtagggtgctggactgtgcccactcccccatgtatctgcccctgcacttaGCTGA GTCCCACTCCAG	272
MPLu10	MPL	A	G	T	A	cds	GE491	AGAGGCTGAGC CATAGACTGT	TGGGCAAGAT TGAAGGTAG	AGAGGCTGAGCCATAGACTGTggtactcagagttctgctgcccctgtcttgccttcaggccctgc cggctccccccagtatcatcaaggccatgggtggagagccaggggaacttcagatcagctggg gaggagccagctccagaaatcagtgattctctgaggtacgaactccgctatggcccagagatcc caagaactcc[a/g]ctggtcccacggtcatacagctgattgccacagaaacctgtgcccctgct ctgcagagccctcactcagctctgctctggaccagttccatgtgctcagcccacaatgccctg gcaagatggaccacaaagcagacctcccccaagtagagaagtatgctgacctcttctgccccacctc tctatctCTACCTTCAATCTTGCCCCA	417
MPLu11	MPL	A	T	D	V	cds	GE490	GGGTTGGAGGC TCTCTCAG	CAGGCTTCCCT AGAGATATTTCT TTTA	GGGTTGGAGGCTCTCTCAGctgacagcagacactagattgtgaagctgggatttctctcccaagg cttcagctctgacagcagaggtggagctgctctcactcagagactccagctggcaactcctac tggctgagctgcgagcgaacctg[a/t]tgagatctctctcctggtgctcctggggtacctggt cctcctgtgactgtggaactgctctggagatgcagggtgagtcacacaaaggaataggagatggg gaggagataAAAGATATCTTAGGGAGCCTG	293
MPLu12	MPL	C	A	A	A	cds	GE482	CTCTGGTGGCA CAATGCCCT	CCAGGATCCC CTGCGTA	CTCTGGTGGCACATGCCCTgtgacagagggacttaagctgctcctctgacatccctgtagt gcgctcccccaaaactgcactgagggagatctccagtgggcatcttggaattggagtggtg agcaccatctgctctggcagc[c/a]caagagacctgttatcaactccgatacacaggagaggg ccatcaggactggaggtatggtcagcaacaaatgcccacagacctcaCTACGAGGGGATCCC TGG	263
MPLu13	MPL	T	A	L	Q	cds	GE450	CTCCCTGCCAA TCCACTG	AAGGATCCAGT ACCAGGCAG	CTCCCTGCCAATCCACTGccatggctcagctgtctctctctctccccccagagactgagggc atgacctgtggcctcactctccagacctgcaccgggtcc[t/a]agggcagtaaccttagggcac tgagcctctgagcccggtgagtgctctctcctcctctgcccacacacacccCTCCCTGGTACT GGATCCTT	203
MPLu2	MPL	C	A	P	Q	cds	GE491	AGAGGCTGAGC CATAGACTGT	TGGGCAAGAT TGAAGGTAG	AGAGGCTGAGCCATAGACTGTggtactcagagttctgctgcccctgtcttgcctcaggccctgc cggctccccccagtatcatcaaggccatgggtggagagccaggggaacttcagatcagctgg gaggag[c/a]agctccagaaatcagtgattctctgaggtacgaactccgctatggccccagag atcccaagaactccactggtcccacggtcatacagctgattgccacagaaacctgtgcccctgct ctgcagaggccctcactcagctctgctctggaccagttctcagctgtgctcagcccacaatgccctg gcaagatggaccacaaagcagacctcccccaagtagagaagtatgctgacctcttctgccccacctc tctatctCTACCTTCAATCTTGCCCCA	417
MPLu3	MPL	T	A	L	H	cds	GE491	AGAGGCTGAGC CATAGACTGT	TGGGCAAGAT TGAAGGTAG	AGAGGCTGAGCCATAGACTGTggtactcagagttctgctgcccctgtcttgcctcaggccctgc cggctccccccagtatcatcaaggccatgggtggagagccaggggaacttcagatcagctgg gaggagccagctccagaaatcagtgattctctgaggtacgaac[t/a]ccgctatggccccagag atcccaagaactccactggtcccacggtcatacagctgattgccacagaaacctgtgcccctgct ctgcagaggccctcactcagctctgctctggaccagttctcagctgtgctcagcccacaatgccctg gcaagatggaccacaaagcagacctcccccaagtagagaagtatgctgacctcttctgccccacctc tctatctCTACCTTCAATCTTGCCCCA	417

FIG. 5YYYYY

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
MPLu4	MPL	C	G	L	V	cds	GE482	CTCTGGTGGCA CAATGCCT	CCAGGGATCCC CTGCGTA	CTCTGGTGGCACAAATGCCTTgTgcacagaaggacttaagctgctccctgctgacatccctgtagt ggcctccccaccccaacttgcactggaggagatctccagtggcat[c/g]tggaattggag tggcagcaccatcgtctctgggcagcccaagagacctgttatcaactccgatacacaggaagg ccatcaggactggagggtatgggtcaagcaacaatgcccacagacctactACCGAGGGATCCC TGG
MPLu5	MPL	G	A	E	K	cds	GE491	AGAGGCTGAGC CATAGACTGT	TGGGGCAAGAT TGAAGGTAG	AGAGGCTGAGCCATAGACTGTggtactcagagttctgagtggccctgtcttgcctcaggccctgc cggctccccccagtatcatcaaggccatgggtggagccagccaggggaaacttcagatcagctgg gaggagccagctccagaaatcagtgatttcttgaggtag[a]aactccgctatggccccagag atcccaagaactccactgggtccacaggtcatcacagtgatggcacagaaacctgctgctctgt ctgcagaggccctcactcagctctgctctggaccagttccatgtctcagcccccaaatggcctg gcaagatggaccaaagcagacctccccaaagtagagaagtatgctgaccttcttctgccccacctc ttatctcTACCTCAATCTTGCCCCA
MPLu6	MPL	C	A	R	R	cds	GE490	GGGTGGAGGC TCTCTCAG	CAGGCTTCCTT AGAGATATTCT TTTA	GGGTGGAGGCTCTCTCAGctgacaggcagacctagattgtgaagctgggatttctccccagg cttcagctctgcagcagaggggtggagctgcctcatctcaggacctccagctgggcaactctac tggctgagctgcg[c/a]agcgaacctgatgggatctcctcgtggctcctgggatactctggt ccctcctgtgactgtggacctggcctggagatgagcaggtgagtcacaagaagaaataggggagatggg gaggagaTAAAGAATATCTCTAGGAAGCCTG
MPLu7	MPL	T	C	F	S	cds	GE472	ACGTGGGGCTG TATCTGACA	CAGGGCTCCCT CTTCCTG	ACGTGGGGCTGTATCTGACaggaacctgagggtctggcctggggaggggattggggccccagcttcc tgaaggaggatgggctaaaggcagcacacagtgggggagagatggcctcctgggccccct[t/ c]catgggtcacctcctcctcctggccccctcaaaacctggcccagtcagcagccaaagtgga ggtgcacagagggtggagatcacctatgcccCAGGAAGAGGGAGCCCTG
MPLu8	MPL	C	T	S	S	cds	GE472	ACGTGGGGCTG TATCTGACA	CAGGGCTCCCT CTTCCTG	ACGTGGGGCTGTATCTGACaggaacctgagggtctggcctggggaggggattggggccccagcttcc tgaaggaggatgggctaaaggcagcacacagtgggcggaagatggcctcctgggcccccttca tggtaacctc[c/t]tgcctcctcctggccccctcaaaacctggcccagtcagcagccaaagtgga ggtgcacagagggtggagatcacctatgcccCAGGAAGAGGGAGCCCTG
MPLu9	MPL	G	A	G	G	cds	GE491	AGAGGCTGAGC CATAGACTGT	TGGGGCAAGAT TGAAGGTAG	AGAGGCTGAGCCATAGACTGTggtactcagagttctgagtggcctgtcttgcctcaggccctgc cggctccccccagtatcatcaaggccatgggtgg[g/a]agccagccaggggaaacttcagatcag ctggaggaggccagctccagaaatcagtgatttcttgaggtacgaactccgctatggccccagag atcccaagaactccactgggtccacaggtcatcacagtgatggcacagaaacctgctgctcctgt ctgcagaggccctcactcagctctctctgctggaccagttccatgtgctcagcccccaaatggcctg gcaagatggaccaaagcagacctccccaaagtagagaagtatgctgaccttcttctgccccacctc ttatctcTACCTCAATCTTGCCCCA

FIG. 5ZZZZZ

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
NGFBd9	NGFB	G	A	A	A	cds	GE1185	CACATCCATAC TGCCTGAGT	AGGGGCCCAGG AGAGTG	CACATCCATACCTGCTCAGTCAGCCCGGGTACGCGTGTGTGCTCCGGTATAACCATGTGCTAGCA CACCCCTTCCCTCTCAGAGAGTGCCCGGTTTGAATGAACCTCTCTGTGATCCCCCTTGGAGGTCA ACTCTGAGGACCCAGAAACTGCTTCTGACTGCACTTAGTACTCCATGAAGTCACTCATTTCT TTTCTCATTCAGGTGATAGCGTAATGTCCATGTGTCTACACTCTGTACAGCTTCTTCTGA TCGGCATAACAGGC(G/A)GAACACACTCAGAGAGCAATGTCTCTGAGGACACACCATCCCCCA AGTCCACTGGACTAAACTTCAGCATTCCTTGACATGCTCTGAGAGCCCTGAGAGCCCGAGCGCCCGG CAGCGGATAGCTGCTGCTGCGGGGAGAGCCCGCAACTTACTGTGAGCCCGGAGCGCTGCTTCT AAAAAGCGGCGACTCGTTCACCCCGTGTGTGTAGCACCCCGCTCCCGTGAAGCTGCAGA CACTCAGGATCTGGACTTCAGGTCGGTGTGTGTGCTGCTCCCTTCAACAGGACTCACAGGAGCAAGC GGTCATCATCCCATCCCATCTTCCACAGGGGCGAATCTCTGCTGTGTGACAGTGTCAAGCTGTGG GTGGGATAAGACCCCGCCAGACATCAAGGCAAGGAGGTGATGGTGTGGAGAGGTGAA CATTAACAACAGTGTATCAACAGTACTTCTTGAGACCAAGTCCCGGAGCCCAATCCCGTTG ACAGCGGTGCGGGGCAATGACTCAAGCACTGGAACCTCATATGTACCAGCACTCAACCTTCT GTCAAGGCGCTGACCATGGATGGCAAGCAGGCTGCTGGCGGTTTATCCGGATAGATACGGCCTG TGTGTGTGTGCTCAGCAGGAAGGCTGTGAGAAAGAGCTGACCTGCCGACACGCTCCCTCCCGCTG CCCCCTCTACACTCTCTCTGGGCCCC
NGFBu1	NGFB	G	C	-	-	noncoding	GE1186	TTTACAGAGGA GCTGACGTTTG	GACAACAGCGG TAACCCG	TTTACAGAGGAGCTGACGTTTGTCTACACATCTACAAGTATGCATAGGAGCTCCCGGAGGCCAGT GAGAGGCCCTCCAGGAGCAGAACTAATCCACAATCTCTGACCAAGTGGGGATATTTGTGG G(G/C)TAAGTGCAGTGCAGTATGGAGTCTCTTGGGACAGTGTAGGACCATACCATTTGATCTA TAGTCACATAAGAAACAACAATAAAGAAAGACATGCTTAAGAGTGAAGAGAAAGGGAGGGA GAAAGAGGAAGGTGGATGGAGGACACTAGCTAGTAAGGGGTCAACTTTGGATCTCTATTCT TGGTTCAGTTTCTATTGTGCTCAGTCTTGTAGTGTAGTCTTCTGTGATCAGTTTCT TTATATGTGAATAAATATGATAAATCTAATGTGACTCACAGCTCATGAGAGATAGAGTGA ACACATTTAAACATCACACAAAGAGGAATATATGTGCTCCCATTTATATATGTGGGTGA GCCTGTGAAGAGTGCCTGGACTAAGATGGTCCAGAGCCCAAGGTTTGTGCAACATGACGC TTGTGAATTCATAACAAGGCTCCAGTCAAGACATCTTCCAGAGTGTCCCTGCTCCCTGCT AAGGGGTACCATCTGTGAGGCTTCAAGACATGCTCCAGCAGATCTTCCCTGCTCCCTGCT GGATTCACAACTGTGTGAGCAGGACGACCATCATCAAGGACAAAGTCCCGGAGGAGGTGT AAACTCTCCCAACCACTCTGTTGACACATGGACACTTACCCTCCCTCAGCGCCTTAA GCTTCAGAGAACTCAAGGACTCTGTAGTGTGCTCCTCAAGCTCATATCGAAGTCTGGGCAAA ATTTCAGGGGCTCTGTCACTCTCTGGAGAGCTCGGATGGGTGACCAACATCCCTACTGCTGT AGTCAGGCCCGGGTTACGCTTACGCTTGTGTC

FIG. 5AAAAAA

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
NGFBu2	NGFB	A	G	-	-	noncoding	GE1186	TTTACAGAGGAGCTGACGTTTG	GACACAGGCG TAACCCG	TTTACAGAGGAGCTGACGTTTGctacacatctacaagatgcataggagctccgaggagccagtgagagccctccaggagcagaactaattccacaattacttgaccaagttgggggattatttgggggtaactgcagtcagtgagtcctctgggacagttagagccataccatttgatctatagtcacataagaacaacaataaaaagaagacatgcttaagagtgaaagagaaaggggagagaaagagaggggtggatggagggacactagcttagtaaggggtcaacttggattctattctgggttcagtttctatttgactcagtccttagtttagttctatttctgtgaatcagtttcccttatagtgaataaataatgataaattcctaatgaactcacagactcatgagaagatagaagtgaaacacatttaaaaaacatcacacaagaggagactattatgtgtccacatttatatatgtgggttagcgtctgaagagggtgctggactaagatgggtccagagccacaggttttgcacacatgacgctttgtgaattcataaagaaggctcc(a/g)agtcaccagatcttagagctgacccagctgactgtctgaaggggtaccagttctgagggctcaagacatgtccccagcagatcttccccgtgcttcccgaggattcaaaaactgtgagcaggagcggcaccatcacatcaagggcacaaagtgccaggagagtggttaacttccccaccacactccctgggtacacacatggacacttacccttccctcagccgcttaagctcagagaaactcaaggactctgttaagtgtgtctccaagctcatatcgaaactactgggcaaaatttcaggggctctgtcacttctctgggagaagctcggtgggtgaccacacacatccatcactgctgagtcagccccCGGTTACGCTGTGTC	1002
NGFBu3	NGFB	G	T	-	-	noncoding	GE1186	TTTACAGAGGAGCTGACGTTTG	GACACAGGCG TAACCCG	TTTACAGAGGAGCTGACGTTTGctacacatctacaagatgcataggagctccgaggagccagtgagagccctccaggagcagaactaattccacaattacttgaccaagttgggggattatttgggggtaactgcagtcagtcagtcctctgggacagttagagccataccatttgatctatagtcacataagaacaacaataaaaagaagaaagacatgcttaagagtgaaagagaaaggggagagaaagagaggggtggatggagggacactagcttagtaaggggtcaacttggattctattctgggttcagtttctatttgactcagtccttagtttagttctatttctgtgaatcagtttcccttatagtgaataaataatgataaattcctaatgaactcacagactcatgagaagatagaagtgaaacacatttaaaaaacatcacacaagaggagactattatgtgtccacatttatatatgtgggttagcgtctgaagagggtgctggactaagatgggtccagagccacaggttttgcacacatgacgctttgtgaattcataaagaaggctcccaagtcaccagatcttagagctgacccagctgactgtctgaaggggtaccagttctgagggctcaagacatgtccccagcagatcttccccgtgcttcccgaggattcaaaaactgtgagcaggagcggcaccatcacatcaagggcacaaagtgccaggagagtggttaacttccccaccacactccctgggtacacacatggacacttaccacctccctcagccgcttaagcttcagagaaactcaaggactctgttaagtgtgtctccaagctcatatcgaaactactgggcaaaatttcaggggctctgtcacttctctgggagaagctcggtgggtgaccacacacatccatcactgctgagtcagccccCGGTTACGCTGTGTC	1002

FIG. 5B BBBB

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
NGFBu4	NGFB	G	A	-	-	noncoding	GE1186	TTTACAGAGGA GCTGACGTTTG	GACAACAGGCG TAACCCG	TTTACAGAGGAGCTGACGTTTGctacacatctcaagtatgcataaggagctccgcggagggccagt gagggccctccagggagcgaactaatccacaattacttgaccaagtgggggattatcttgagg ggtaactgcagtcgagtatggagtcctcttgggacaggttagagccataccattgatctatagt cacataagaacaacaataaaaagaaagacatgcttaagagtgaaggaagggagggagaaa agaaggaaagggaggatggaaggagacacagcttagtaagggtgcaacttggaattctattctggg tcagttttcatttgtagctcagtcgcttagtgtagttcatttctggaatcagttccctac atgtgaataaatatgataaatcctaattgaactcacagacatcatgagaagtgaagtgaacac atttcaaaaaacatcacacaagaggaaactattatgtggtccacatttatatgtgggttagcgt ctgaagggtgcttgactaagatggctccagagccacaagggtttttgccaacatgacgctttg tgaattcataaagggctccaagtcaccagatcttagagctgacccagtcactgtctgaaaagg gggtaccagttctgagggctccaagacatgtcccagcagatctccccgtgcttcccagaggat tcaaaactgttgagcaggcggcaccatcacatcaaggcacaggtgccaggagaglg/a tgtt aaacttcccccaaacctcccgtgtacacacatggacactaccaccccctcagcgcctaa gcttcagagaaactcaaaaggactctgaagtga gtctccaagctcatatcgaaactactgggcaaa atttcagggggtctgtcacttcttgagaagctcggatgggtgaccacacatccatctgctg agtcagccccGGGTACGCTTGTC
NGFBu5	NGFB	T	C	V	A	cds	GE1185	CACATCCATAC TGCCTGAGT	AGGGGCCCCAGG AGAGTG	CACATCCATACTGCCCTGAGTcagccccgggttacgacctgtgtgccgggtataaaccattgctagca caccccttccccctcagaaagtcccccggtttgaatgaacctcttcgtgatccccctggagggtca actctgagggagcccaagaaactgcctttgactgcatttagactccatgaagtcacctcatcttc ttttcattccaggtgcatagcgtaatgtccatgtgtttcacactctgatcacagctttctga tcggcatacagcgggaaccacatcagagagcaatgtccctcaggagacacacatccccaaag t /c ccactggactaaactcaagcatccccctgacactgacctgcagagcccgccagcgcgccgg cagcggcgatagctgcacgctggggggcagaccgccacatctactgagccccaggtctgtt aaaaagcgggactccgttcaacccctgtgctgttttagacaccagcctccccgtgaagctgcaga cactcaggatcttgacttcgaggtcgggtggtctgcccccttcaacaggactcacaggagcaagc ggctcatatccatccccatttccacagggcggaattctcgtgtgtgtgagtcagtcgctggtg gttggggataaagaccacccacagacatcaagggcaagggtgatggtgttgggagaggtgaa catcaacaacagtgatcaaacagtaactttttgagacaaagtgcgggacccaatccccgtg acagcgggtgcgggggcatgtactcaaaagcactggaactcatattgtaccacgactcacacctt gtcaaggcgctgaccatggatggcaagcaggctgcctggcggtttatccggatagatcagggcctg tgtgtgtgtcagcaggaaggctgtgagaagaagcctgactgcgcagcagctccctccccctg cccttctcaCTCTCTCTGGGCCCCCT

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
NGFBU6	NGFB	T	C	-	-	noncoding	GE1186	TTTACAGAGGA GCTGACGTTTG	GACAACAGCGG TAACCCG	TTTACAGAGGAGCTGACGTTTGCTACACATCTACAAGTATGATAGGAGTCCGCGGAGGCCAGT GAGAGGCCCTCCAGGAGCAGAGCACTAATCCACAATTAC(t/c)TGACCAAGTTGGGGATTATTT GTGGGTAACTGCGAGTGCAGTATGAGTCCCTTGGGACAGTTAGAGCCATACCATTGATCTA TAGTCACATAAGAACAAACAATAAAGAAAGACATGCTTAAGAGTGAAGAGAAAGGAGGGA GAAAGAGGAGGTTGATGGAAGACACTAGCTTAGAGGGTCAACTTCTGATCTCTATTC TGGTTCAGTTTCTATTTGACTTCAGTGTCTTAGTGTAGTCTTCTGTGAATCAGTCTTC TTATATGTGAATAAATATGATAAATCTAATTGAATCAGACATCATGAGAGATAGAGTGA ACACATTTTAAACATCACACAAGAGGAACTATTATGTGGTCCACATTTATATGTGGGTGTA GCCTCTGAAGGGTCCCTGGACTAAGATGGTCCAGAGCCACAAGTTTTCACAACATGACGC TTGTGAATTCATAAAGGGCTCAAGTCCAGATCTAGAGTGAACCATGCACTGCTGA AAGGGGTACCACTCTGAGGCTTCAAGACATGTCCTCCAGCAGATCTTCCCGTGCCTCCAGA GGATCAAACTGTGAGCAGGAGGACCATCATCAAGTGAAGTGAAGTGAAGGAGGAGTGT AACTCTCCCAACCACTCCCTGGTACACACATGGACATTCACCTCCCTCAGCGCCTTAA GCTTCAGAGAACTCAAGGACTCTGTAAGTGTCTCAAGCTCATATCGAATCTACTGGGCAAA ATTCAGGGGCTCTGCTCACTCTCTGGAGAGAGTCTGGATGGGTGACCAACATCCCATCTGCTG AGTCAGCCCCGGGTACCCCTGTTC	1002
NGFBU7	NGFB	C	T	-	-	noncoding	GE1186	TTTACAGAGGA GCTGACGTTTG	GACAACAGCGG TAACCCG	TTTACAGAGGAGCTGACGTTTGCTACACATCTACAAGTATGATAGGAGTCCGCGGAGGCCAGT GAGAGGCCCTCCAGGAGCAGAGCACTAATCCACAATTACTTGACCAAGTTGGGGATTATTTGGG GGTAAGTGCAGTGCAGTATGAGTCTCTTGGGACAGTGAAGCCATACCATTGATCTATAGT CACATGAAGAACAAACAATAAAGAAAGACATGCTTAAGAGTGAAGAGGAGGAGGAGGAAA AGAAGGAAGGTTGGATGGAAGACACTAGCTTAGTAAAGGGTCAACTTGGATCTCTCTGGT TCAGTTTCTATTTGAGTCTCAGTCTTCTAGTGTAGTCTTCTGTGAATCAGTCTCTCTAT ATGTGAATAAATATGATAAATCTAATTGAATCAGACATCATGAGAGATAGAGTGAACAC ATTTAAACACATCACACAAGAGGAACTATTATGTGGTCCACATTTATATGTGGGTAGCGT CTGAAGAGGTGCTGGACTAAGATGGTCCAGAGCCACAAGTTTTCACAACATGACGCTTTG TGAATTCATAAAGGGCTCAAGTCAAGACATCTTCCAGCAGATCTTCCCTGCTTCCAGA GGTACCAGTTCTGAGGCTTCAAGACATGTCCTCCAGCAGATCTTCCCTGCTTCCAGA GGATCAAACTGTGAGCAGGAGGACCATCATCATCAAGTGAAGTGAAGTGAAGGAGGAGTGT AACTCTCCCAACCACTCCCTGGTACACAATGGACATTCACCTCCCTCAGCGCCTTAA GCTTCAGAGAACTCAAGGACTCTGTAAGTGTCTCAAGCTCATATCGAATCTACTGGGCAAA ATTCAGGGGCTCTGCTCACTCTCTGGAGAGAGTCTGGATGGGTGACCAACATCCCATCTGCTG AGTCAGCCCCGGGTACCCCTGTTC	1002

FIG. 5DDDDDD

[illegible]

FIG. 5EEEEE

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
NTRK1d10	NTRK1	C	T	-	-	noncoding	GE1290	GCTGCCCTGGG TGAACA	GGGATGCTCTAT AGGGAAGGGA	GCTGCCCTGGGTGAACAGcagtgagggt(c/t)ggccccaaactcagtcctcgtccctgccgcttc 320 catccaggcactgaaggagggtccgagagtgctcggcaggactccaacgtgaggctgagctgc tcaacatgctgcagcaccagacacatcgtcgcttcttcgggctcgcacgggggccccctg ctcatggcttcgagtatatcgggcacgggacctaaccgcttctccggtaccagaccctggc ctacagcgtggccccggccccctggtctctggccccgtctTCCCTTCCCTATAGACATCCC
NTRK1d9	NTRK1	G	C	-	-	noncoding	GE1067	GGAGGCTCTGA GAGTACAGGAG	AGTAGGAAACA AAGCCAGGAG	GGAGGCTCTGAGAGTACAGGAG(g/c)agccccggatctaaactaccctgtccccaccaggctc 223 tgggtggctgggctggcctggcgtcttggcggctctctcttctacgctgctcttggctcaa caaatgtggacggagaaacaagtttgggatacaaccgtagtcgggctgcagagggtgctgtctc gctgtctTCTGGCTTGTTCCTACT
NTRK1u1	NTRK1	A	G	Q	Q	cds	GE1290	GCTGCCCTGGG TGAACA	GGGATGCTCTAT AGGGAAGGGA	GCTGCCCTGGGTGAACAGcagtgagggtcgcccccaactcagtcctcgtccctgccgttccatc 320 caggcactgaaggagggtccgagagtgctcggcaggactccaal(a/g)cgtaggctgagctgc tcaccatgctgcagcaccagacacatcgtcgcttcttcgggctcgcacgggggccccctg ctcatggcttcgagtatatcgggcacgggacctaaccgcttctccggtaccagaccctggc ctacagcgtggccccggccccctggtctctggccccgtctTCCCTTCCCTATAGACATCCC
NTRK1u2	NTRK1	C	T	F	F	cds	GE1290	GCTGCCCTGGG TGAACA	GGGATGCTCTAT AGGGAAGGGA	GCTGCCCTGGGTGAACAGcagtgagggtcgcccccaactcagtcctcgtccctgccgttccatc 320 caggcactgaaggagggtccgagagtgctcggcaggactccaacgtgaggctgagctgctcac catgctgcagcaccagacacatcgtcgcttcttcgggctcgcacgggggccccctg ctcatggcttcgagtatatcgggcacgggacctaaccgcttctccggtaccagaccctggc ctacagcgtggccccggccccctggtctctggccccgtctTCCCTTCCCTATAGACATCCC
NTRK1u3	NTRK1	C	T	H	Y	cds	GE1129	CAGGCTCCTGG GAGTTCTATC	CCAGGGTGTCT ACAGTTTGGAT	CAGGCTCCTGGGAGTTCTATCctccagcctatccccctctcttcttctgttccagatccc(c/t)atggaccgatgcgaagctgctggctgggtgggaggatgtggctccaggccccctgggtctggg ggaagctgctggcctggctagccaggtcgctgcgggagtggtgtaacctggcggtctgcattttg tgcacgggacctggccacacgaactgtctagtggccagggaactgggtggccaagattggatgat ttggcatgagcagggtatctacagcaccgactattaccgtgtaagggtccttctgtccccaaag cctccccctgcATCCAAACTGTAGACACCTGG
NTRK1u4	NTRK1	C	T	A	A	cds	GE1129	CAGGCTCCTGG GAGTTCTATC	CCAGGGTGTCT ACAGTTTGGAT	CAGGCTCCTGGGAGTTCTATCctccagcctatccccctctcttcttctgttccagatccccatg 358 gaccgatgcgaagctgctggctgggtgggaggatgtggctccaggccccctgggtctggggcag ctgctggc(c/t)gtggtagccaggtcgctgcgggagtggtgtaacctggcggtctgcattttg tgcacgggacctggccacacgaactgtctagtggccagggaactgggtggccaagattggatgat ttggcatgagcagggtatctacagcaccgactattaccgtgtaagggtccttctgtccccaaag cctccccctgcATCCAAACTGTAGACACCTGG
NTRK1u5	NTRK1	T	G	-	-	noncoding	GE1067	GGAGGCTCTGA GAGTACAGGAG	AGTAGGAAACA AAGCCAGGAG	GGAGGCTCTGAGAGTACAGGAGggccccctggatctaactaccctgtccccaccaggctcgg 223 tggctgtgggctggcctggcgtcttggcgtgctctctcttctacgctgctcttggctcaaaaaa tgtggacgggaaacaagtttgggatacaaccg(t/g)agtcggggtgcagagggtgctgtctct gctgtctTCTGGCTTGTTCCTACT
NTRK1u6	NTRK1	C	T	G	G	cds	GE1129	CAGGCTCCTGG GAGTTCTATC	CCAGGGTGTCT ACAGTTTGGAT	CAGGCTCCTGGGAGTTCTATCctccagcctatccccctctcttcttctgttccagatccccatg 358 gaccgatgcgaagctgctgggtgggtgggaggatgtggctccagg(c/t)ccccgggtctggg ggaagctgctggcctggctagccaggtcgctgcgggagtggtgtaacctggcggtctgcattttg tgcacgggacctggccacacgaactgtctagtggccagggaactgggtggccaagattggatgat ttggcatgagcagggtatctacagcaccgactattaccgtgtaagggtccttctgtccccaaag cctccccctgcATCCAAACTGTAGACACCTGG

FIG. 5FFFFF

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
NTRK1u 7	NTRK1	G	T	G	V	cds	GE1129	CAGGCTCCTGG GAGTCTATC	CCAGGCTGTCT ACAGTTTGGAT	CAGGCTCCTGGAGTTCTATCTCccagcctatccccctctctcttcttctgttcacagatccccatg gacccgatgccaagctgctggtg/g/tl-999gagatggtggtccagggccccctgggtctggg gcagctgctggcgtggtgtagcaggtgctgcggtggtggtacccctggggtctgcatcttg tgcacccggacccctggccacacgacgtctctagtggccagggactggtggtcaagatggtgat tttggcatgagcaggatattctacagcaccgactataccgtgtaagggtcccttctcccccaag cctccccctgCATCAAACTGTAGACACCCCTGG	358
NTRK1u 8	NTRK1	C	T	F	F	cds	GE1042	CCAGGGGCCCG ACTAGC	AGCCCAAGTCT GGGTCTCTCT	CCAGGGCCCGAGTAGCTgagacctggggactgatcctcctgeacccccccccagaccatcgctg aagagtgtctctcgtttt(c/t)gtggcgccagatgcttccatttcactcctcggtcagtcgcc tgtgagtgtggccagtgtgctggcagtggtggagltggggAGGACACCCAGACTTGGGCT	187
PACEd1 2	PACE	g	T	-	-	noncoding	GE1219	GAGTCGCAGGG GGTGCC	AGCAAGGCCAA GCACGC	GAGTCGCAGGGGTGCCcctggtccctctggggcaggctgacccatcatggtgctcctctgcaca gagacatcgggaaacggctcgaggtgcggaagaccgtgacccggtgctggggcagagcccaaccac atcactcggtggagcacgctcagggcggtcaccctgtctctataatcgccgtggcagcctggc catccactggtcagccccatgggcacccgtccacctgctggcagccaggtgcttctctgtc cctggccgccccctggcagcgccgctg/g/l cctctcacagcgcctgctgcttgccttgcct	320
PACEd1 3	PACE	G	T	R	R	cds	GE1219	GAGTCGCAGGG GGTGCC	AGCAAGGCCAA GCACGC	GAGTCGCAGGGGTGCCcctggtccctctggggcaggctgacccatcatggtgctcctctgcaca gagacatcgggaaacg/g/t ctcgaggtgcggaagaccgtgacccggtgctggcgagcccaaa ccacatcactcggtggagcacgctcagggcggtcaccctgtcctataatcgccgtggcgagccac tggccatccactggtcagccccatgggcacccgtccacctgctggcagccaggtgcttgcctc tgtctctgccccctgccccagcgccgctctctctcacagcccGGTGTGCTTGGCT	320
PACEu1	PACE	C	G	G	G	cds	GE687	GTCTGTTTACG TGACACACACT T	CAGCTGGGTC CMAACC	GTCTGTTTACGTGACACACACTTgccccctctctccacgcggcagtggtcgaggaaggctctctc ctgcacagaagagctgtgtccagactgccccctcagg/g t ctgcccccccaagctctctcgata cgactatagcacagagaatgagctggagacacatccggcgagcgtctgccccctgcccacgct tcagtggccacatgcccagggccgccccctgacagactgctcagctgccccagccagcctctt ggacctgtggagcagacttgcctcccggaaggccagagcagcagagtcctcccgccacacagcgc agccacctcggtgcccccgaggtggagggcggggcaacggctgccccagggctgctgccccca cactgctctgagtggtggcgccctcagctgccttcactgctggtctctctcactgctctt cctggtcctgcagctgcctctggtcttagtttctggggggtgaaagtgtacacctggaccgtg gctcatctctcacaaagggtgccccctgagcctggagcctggcagagaggtgccccctgactcaga gaggacaggccggcgagagagacgcttatacaagaccagagcgccccctctgctgagcaca ctgccccccccctcaagcctccccctctctggcacttttataatcaccaagatcttcttat cttgggactggTTTGGACCCCGCTG	742
PACEu1 0	PACE	G	A	G	S	cds	GE548	GAGAGCTGCTC CCAAAA	ACCTAACTCCA CATCCAGCC	GAGAGCTGCTCCAAAAAagactgatccccagcctctctctcttcttctgagtaagaacctca catggcgggacatgcaacacctggtggtacagacctcgagccagccacctcaatgccaacgac tgggcccacccaat/g/a gtgtggcggaaggtagggcgaggtgccccggcagGGCTGGATGTG GAGTTAGGT	204
PACEu1 1	PACE	T	A	L	Q	cds	GE1219	GAGTCGCAGGG GGTGCC	AGCAAGGCCAA GCACGC	GAGTCGCAGGGGTGCCcctggtccctctggggcaggctgacccatcatggtgctcctctgcaca gagacatcgggaaacggctcgaggtgcggaagaccgtgacccggtgctggcgagcccaaccac atcactcggtggagcacgctcagggcggtcaccctgtctctataatcgccgtggcgagccacac catccacct/a ggtcagccccatgggcacccgtccacctgctggcagccaggtgcttgcctc tgtccccctgccccctgccccagcgccctctctctcacagccccGGTGTGCTTGGCT	320

FIG. 5GGGGGG

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
PACEu2	PACE	G	C	S	T	cds	GE687	GTCTGTTTAGC TGACACACACT T	CAGCTGGGGTC CANACC	GTCTGTTTAGCTGACACACACTTgccccctctccacgcccggcagtggtgcgaggaagggtctctcc ctgcaccagaagagctgtgtccagcactgccccctccaggtctgcgcccccaagtcctctcgatacgc ctatagcaccgagaaatgacgtgagagaccatccggccagcgtctgcgccccctgcacacgtctcat gtgccacatgccaggccggccgctgacagactgctcagctgccccccagccacgctctcttgac cctgtggagcagacttgctcccgcaagcagagcagcagcagcagcagcagcagcagcagcagcagc acctcggtgccccccggagggtgagggcgggcagcagcagcagcagcagcagcagcagcagcagc tgccctgaggtgtgtggcgccctcagctgcgcttcacgtggtggtctctcactgctctctctg gtccctgagctgcctgtggtcttcttctgggggtgaagggtacacacagcagcagcagcagcagc catctctacaagggtgccccctgaagcctggcagggagggagcagcagcagcagcagcagcagc acgagggccggggcgagagagcagcagcagcagcagcagcagcagcagcagcagcagcagcagc ctgcccccccccaagcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagc cttgggagctgGGTTTGGACCCCGAGCTG
PACEu3	PACE	C	T	-	-	noncoding	GE590	GTCCACAGGC CGAGGG	AGCCCCATCTC CCCAGC	GTCCACAGGCGGAGGGGggtgggtccatcttctgtctgggc[c/t]tcggggaaacggggcgg ggaacatgacagctgcaactgcagcagcgtgacacacacagtatctacacgtgtccatcagcagcg ccacgagcttggcaacgtgcgtggtagagcagcagcagcagcagcagcagcagcagcagcagc agcagtggaacacagaaatgagaagcagatcgtgagctctacactgggggGCTCGGGGAGATG GGGCT
PACEu4	PACE	G	C	L	L	cds	GE1219	GAGTCGAGGG GGTGCC	AGCAAAAGGCAA GCACCC	GAGTCGAGGGGGTCCCGctgggtccctctgggcccaggtgaccatcatggtgctctctctgcaca gagacatcggaacacggctcgaggtgcggaagacgtgacgcggtgcttgc[c]ggcgagcccaa ccacatcactcgtggagcagctcagcagcagcagcagcagcagcagcagcagcagcagcagc tggecatccactggtcagccccatgggcaacccgtccacactgctggcagcagcagcagcagc tgctcctgccccctgccccagcgcgctctctcagagccccGGTGTGCTTGGCT
PACEu5	PACE	C	T	G	G	cds	GE1219	GAGTCGAGGG GGTGCC	AGCAAAAGGCAA GCACGC	GAGTCGAGGGGGTCCCGctgggtccctctgggcccaggtgaccatcatggtgctctctctgcaca gagacatcggaacacggctcgaggtgcggaagacgtgacgcggtgcttgc[c]ggcgagcccaa ccacatcactcgtggagcagctcagcagcagcagcagcagcagcagcagcagcagcagcagc tggecatccactggtcagccccatgggcaacccgtccacactgctggcagcagcagcagcagc tgctcctgccccctgccccagcgcgctctctcagagccccGGTGTGCTTGGCT
PACEu6	PACE	T	C	-	-	noncoding	GE1195	AGCCAAATCCCT TCCTTGG	CAGCATCGGGT GGGAGG	AGCCAAATCCCTTCCTTGGcactttttaaatacacaagatttttattcttgggactgggttt ggacccccagctgggaggaagaggggtggagactg[c/t]ctcccatcctacctcgggccccacc tgccacacctgaggtggccccagcagcagcagcagcagcagcagcagcagcagcagcagcagc cccttccatgtggagaaaggagtgaaacctttaggcagcttgccccggccccccagccag agttcctgcggaagtgaaggaggagcagcagcagcagcagcagcagcagcagcagcagcagc tgccccctccttcccttaagcaataatggtcccaaccagcagcagcagcagcagcagcagcagc atatctgagggaggagggcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagc gtgagctctggcgaggcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagc cagtggtcagctggcctgtgctgtgtccatgacccacccctgctcagcagcagcagcagcagcagc actggccacacaggtggcgagcagcagcagcagcagcagcagcagcagcagcagcagcagcagc caccctccccctctcttgcaacccgctctcctcagggcccaagtcctgtgtgtgtgtgtgtgtgt ctgctggggt ccagccgggct

FIG. 5HHHHHHH

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
PACEu7	PACE	G	A	-	-	noncoding	GE195	AGCCAATCCCC TCCTTGG	CAGCATCGGGT GGGAGG	AGCCAATCCCCCTCTGGGcaactttttaaattcaccaaagtatttttttatcttgggactgggttttggacccagctgggggcaaggaggggtggagactgtttcccatctaccctcgggccacacctggccacctgaggtggggccaggaccagctgggggaggggagggcggtacccacacctcagacaccttccatgtggagaaaggagtgaaccttttagggcagcttgcctggggcccccagccagcagagttcttgcggagtgaagggggcagccctgtgtgtggatctctgacccagggcccccagcagctcttggcccttcccttcccttaagcaataatgggtcccatcag[g/a]cagtcgggggctggcctaggagataatctgagggagggccacctctccaaaggctctcgcacctccacacctgtcccccagctctgtgtgagcttggggcagcagccatcataggaagggaaggaaggagggaggtgctcaggtgtgcacgtggcatgtggcctgtggcctgtgtccatgacccacctgtgtcctcgtgcctccacacctactggccacagctggcgagcaagggcgaagctctgtgtaacctgtgtgtggtgtcctgacacacctccctctctgacacccctctccgtcagggcccaagctccctgtttctgagccccggctgctggctgtgtggcactcacagacctggagccccctgggtgggtgggtggggaggggctggcctggccagccggcctctctggCCTCCACCCGATGCTG	814
PACEu8	PACE	C	T	-	-	noncoding	GE195	AGCCAATCCCC TCCTTGG	CAGCATCGGGT GGGAGG	AGCCAATCCCCCTCTGGGcaactttttaaattcaccaaagtatttttttatcttgggactgggttttggacccagctgggggcaaggaggggtggagactgtttcccatctaccctcgggccacacctggccacctgaggtggggccaggaccagctgggggagggcggtacccacacctcagacaccttccatgtggagaaaggagtgaaccttttagggcagcttgcctggggcccccagccagcagagttcttgcggagtgaagggggcagccctgtgtgtggatctctgacccagggcccccagcagctcttggcccttcccttcccttaagcaataatgggtcccatcagggcagctgggggctggcctaggagatatctgagggagggaccctctccaaaggctctcgcacctccacacctgtcccccagctctgtgtgagtggtggcggcagcagccatcataggaagggaaggaaggagggaggtgctcctccagctgtgcacgtggcctgtggcctgtgtccatgacccacctgtgtcctcgtgcctccacaccttggcatgtggcctgtggcctgtgtccatgacccacctgtgtcctgacacacctggccacagctggcgagcaaggg[c/t]cgaagctctggctgaacctgtgtgtgtgacgacacctccctctctgacacccctctccgtcagggcccaagctccctgtttctgagccccggctgctggctgtgtggcactcacagacctggagccccctgggtgggtgggtggggaggggctggcctggccagccggcctctctggCCTCCACCCGATGCTG	814
PACEu9	PACE	G	C	L	L	cds	GE590	GTCCACAGGGC CGAGGG	AGCCCCATCTC CCCAGC	GTCCACAGGGCCGAGGGgggctgggctccatcttctgtctgggctctggggaacggggccgggaaatgacagctgcaactgcagcggctacacaaacagtatctacagct[g/c]tccatcagcagcgccacgcagtttggaacagtgccgtggtaacgcgagggcctgtgttccacactggccacagacctacagcagtggaaccagaatgagaagcagatcgtgagtttacctgggggtggggGCTGGGAGATG GGGCT	265
PA11d10	PA11	C	T	-	-	noncoding	GE285	GGGAGACCCCTG TCCTTAAGAA	GTCTGCATCTG TCGGACTA	GGGAGACCTGTCTTAAGAAaacggcgggggtgggggtgggtggcagtgccagatccctctgttctaaagacattgtcctctctctgtcagctgtcatagctcagcccgatggccccggaggagatc atcatggacagaccttccctcttctgtgtgtggcgacacacccacaggtgagcctggaaacctca [c/t]gttccacatctccacaccttcttctctcagggaactAGTCCCGACAGATGCAGAC	257
PA11d11	PA11	G	A	-	-	noncoding	GE384	AGAAGAGCAGG GCCACAG	CCAGCCTTTCC TTGCTTC	AGAAGAGCAGGGCCACAGctggcagctccctccctccc[c/g/a]cctgtcttccagaacgat tccctccacagcccttctccatgtctctaggatgagatgtctccagccctcacctgcttagtc ctgggcttggcccttcttctgtggaagggtctgtgtgacacctccccctcctacgtggccca cctggcctcagactcgggtgaggggtgttccagcaggtggcgagccctccaaaggaccgcaacg tgggttctccacctatgggtggcctcgtgtgtggcctatgtccagctgacacacagaggaggaa acccagcagcagattcaagcagctatgggttcaagattgattgagcccggaacaccagggg aggtgggtggcatgcagaaacagacctaccaGAAGCCCAAGGAAGGCTGG	439

FIG. 5IIIII

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
PA11u1	PA11	T	G	S	A	cds	GE368	GCCCTAAGAGGAAAGGAA	GGGTCTGGGA CCAGTTATC	GCCCTAAGAGGAAAGGAAacctctcttgcagagcggcagatcctaactctgtatccacatctgttcgagaccagagcctctccacgtcgcgcagcgtgcgagaaagtcgaagatcgcaggtgaacgagagtcgcacgtggcctctca [t/g]ccacaggtgagcttggctcaggtgaggtccacgggtgtcgccctccatcgcccttcagGATAACTGTTCCCCAGACCC
PA11u2	PA11	G	A	V	I	cds	GE384	AGAAGAGCAGG GCCACAG	CCAGCCTTTCC TTGGCTTC	AGAAGAGCAGGCCACAGctggcagctccctccctcccgctgtctccagaacgattccttcacagccctcttccatgtctcagatgcagatgtctccagcctccactgcctgctcctggacctg [g/a]tcttggtagaggtctgtctgcacatccccatctcactcgttggcccacttggtctccagacttcgggtgaggtgttccagcaggtggcgcagcctccaaagaccgcaacgttgttctcaccctatgggtggcctcggttgcctccagctccagctgacacagggaggagaaacccagcagcagattcaagcagctatgggattcaagattgattggtgagccacgggacacccagggagggtgggtggcctgcagacagacctaccgaGAGCAAGGAAGGCTGG
PA11u3	PA11	G	A	A	A	cds	GE379	GGTAGCCCCCA TCCATCAG	CAACTGGGATA TGTGGATTAT TT	GGTAGCCCCCATCCATCAGagtgagcctctgtgggggaagtgggctcggtcggcggaaacctcaattcagcataagcctcacatgtcctctctctctgtctccgggtgcagacaaggcatggccccgccctcgcgcactctgacaaggagctcagggccatggaacaagatgagatcagcacacagacgc [g/a]atctctgtccagcgggtcgaagctgggtccagggcttcagccccactcttcagggtgttccggagcaggtcaagcaagtggacttccagaggtggagagagcagattcattcatcaatgactgggtgaagacacacaaaaggtgagcagggaaggaaacccatttctctgggctcgaagagaaagggaatttggAAATAATCCACATATCCCACTG
PA11u4	PA11	G	A	A	T	cds	GE384	AGAAGAGCAGG GCCACAG	CCAGCCTTTCC TTGGCTTC	AGAAGAGCAGGCCACAGctggcagctccctccctcccgctgttctccagaacgattccttcacagccctcttccatgtctcagatgcagatgtctccagcctccactgcctgctcctggacctg [g/a]ccttgtcttggtagaagggtctgtctgcacatccccatctcactcgttggcccaactggcctcagacttcgggtgaggtgttccagcaggtggcgcagcctccaaagaccgcaacgttgttctcaccctatgggtggcctcggtgttcgacctccagctgacacagggaggagaaacccagcagcagattcaagcagctatgggattcaagattgattggtgagccacgggacacccagggagggtgggtggcctgcagacagacctaccgaGAGCAAGGAAGGCTGG
PA11u5	PA11	A	G	D	G	cds	GE379	GGTAGCCCCCA TCCATCAG	CAACTGGGATA TGTGGATTAT TT	GGTAGCCCCCATCCATCAGagtgagcctctgtgggggaagtgggctcggtcggcggaaacctcaattcagcataagcctcacatgtcctctctctctgtctccgggtgcagacaaggcatggccccgccctcgcgcactctgacaaggactcagggccatgggaacaag [a/g]tgagatcagcaccacagacgcatctctgtccagcgggtatctgaagctgggtccagggctcatgccccacttcttcagggtgttccggagcaggtcacaagcaagtggacttccagaggtggagagaccagattcattcatcaatgactgggtgaagacacacaaaaggtgagcaggcagggaaggaaacccatttctctgggctcgaagagaaagggaatttggAAATAATCCACATATCCCACTG
PA11u6	PA11	G	C	G	R	cds	GE363	AAC TAGGTGAA AACCCAGTGCT	GTCTGCAACC GTACACAGAT	AAC TAGGTGAAAACCCAGTGTgggttgccatcagaagagctggagccatttccaaagaccatctgtcgtcttcacagctgagtccaccagccccatggccattactacgacatcctggactgccctaccac [g/c]gggacacctcagcatgttcatgtgtccccctatgaaaagagggtgctctcttgcctcaccacattctgagtgccagctcatcagcactggaaagcaacatgaccaggctgccccctctctgggtcttgccccaaagtaagccaccccgctatctccccgacctaccacccctctcttngctccctaaagtcacgcccccgaggttcaatttcccaatctctgacgcttTGCAAGCTTGCAAGCA

FIG. 5JJJJJ

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
PA11u7	PA11	A	T	I	F	cds	GE379	GCTAGCCCOCA TCCATCAG	CNACTGGGATA TGTGGATTTAT TT	GCTAGCCCCCATCCATCAGAGtgaggcccttgtgagggaagtgtgggggaagtgtggctcggctgggaaccttcaa ttcagcatagccctcacatgtctctctctctctgtcctcgtgcagacaaaggtgagatcagcaccacagacgc ctccggcatctgtacaaggagctcatgggcatggaacaaaggtgagatcagcaccacagacgc g/a/t tcttcgtccagcgggacttgaagctgggtccagggtctcatgccactcttctcaggctg ttccggagcacgggtcaagcaagtggactttcagaggtggagagagccagattcatcatcaatga ctgggtgagacacacaaaaggtgagcagggaggaagaaagaaacccatttcttgggacctcaag agaaagggaattctggAAATAATCCACATATCCCATGTG
PA11u8	PA11	A	T	L	L	cds	GE298	GTAGGGGATGG GGAAAGGT	TCTCAAGGAGA GGTTCTTTTTT	GTAGGGGATGGGAAAGGTggagctgccagccagaggggagcccggtctgagcagaccttcttgc tgcatactgcagggtctccctggagactgaagtgcagctcaggaagccctt a/t gagaacctg ggaatgaccgacatgttcagacagtttcagagtgacttcagagcttctcaggttaagaagacttt cccttgcatttctcaccctcagtgagctgcggggggccccctaaagggAAAAAGGAACCTCTCTCTTG AGA
PA11u9	PA11	A	T	-	-	noncoding	GE285	GGGAGACCCCTG TCTCTAAGAA	GTCTGCATCTG TCGGGACTA	GGGAGACCCCTGTCTCTAAGAAaaaacggcggggggtgggggtgggtgccagtgccagcatccctctgt tctaagacattgtccctctctctgc a/t tgctgctcatgtctcagccgcatggcccccgagga gatcatcatggacagaccttctctt tggttcggcacaacacacacaggtgagcctggaaaccc atcacgttccacatctccacccattcttctctcaggaactTAGTCCGACAGATGCAGAC
PA12d1 5	PA12	C	T	-	-	noncoding	GE341	TCTACTGTGTG CTGACCTCAC	GCATAACAAG AGCAGTCTCC A	TCTACTGTGTGACTCACccaaaatgttaccttatgtttcagagctgttttttcttctctct cttgcctctagattgaacaaatggaggatctttgtgtggcaaacacactcttggccctcaattt attcaagcatctggcaaaagcaagcccccacacagaaaccttctctctccccatggagcatctcgt ccacctggccatggtctacatgggtccaggggcagaccgaaagccagatggccaaggtgagt ccacctggccatggtctacatgggtccaggggcagaccgaaagccagatggccaaggtgagt ttgagctgaagctccacatttgggc c/t gagtagtctctgaatggaaatTGGAGAACTGCTCTT GTTATGC
PA12d1 6	PA12	T	G	-	-	noncoding	GE341	TCTACTGTGTG CTGACCTCAC	GCATAACAAG AGCAGTCTCC A	TCTACTGTGTGACTCACccaaaatgttaccttatgtttcagagctgttttttcttctctct cttgcctctagattgaacaaatggaggatctttgtgtggcaaacacactcttggccctcaattt attcaagcatctggcaaaagcaagcccccacacagaaaccttctctctccccatggagcatctcgt ccacctggccatggtctacatgggtccaggggcagaccgaaagccagatggccaaggtgagt ttgagctgaagctccacatttgggcgag t/g agtctctgaatggaaatTGGAGAACTGCTCTT GTTATGC
PA12d1 7	PA12	C	G	-	-	noncoding	GE341	TCTACTGTGTG CTGACCTCAC	GCATAACAAG AGCAGTCTCC A	TCTACTGTGTGACTCACccaaaatgttaccttatgtttcagagctgttttttcttctct c /g tcttctgtctcagattgaacaaatggaggatctttgtgtggcaaacacactcttggccctca attattcagcatctggcaaaagcaagcccccacacacacacacactcttctctccccatggagcatc tcgtccacctggccatggtctacatgggtccaggggcagaccgaaagccagatggccaaggt gagttgagctgaagctccacatttgggcgagtagtctctgaatggaaatTGGAGAACTGCTCTT GTTATGC
PA12d1 8	PA12	G	A	-	-	noncoding	GE318	CCACTGATAG CCATCTGTTTT	CCAGTCTGACA CTGTGGCTTTA	CCACTGATAGCCATCTGTTTTTaaagttcagtaaatccgtgggcaagtgtaacc g/a ttcttc tctaattctgattgcaggtgctcagtttaataagagtgaggagcaatgcagttaccctccagact ccagagaaatttaccagctggtggtctcatgcagcagatccagaaaggttagttatctctgatgcgat tttgcaggtatctgactactggtccaaaattcttcttgggtttatttttgcgaattctctctct TAAAGCCACAGTGTACACTGG
PA12d1 9	PA12	C	A	N	K	cds	GE318	CCACTGATAG CCATCTGTTTT	CCAGTCTGACA CTGTGGCTTTA	CCACTGATAGCCATCTGTTTTTaaagttcagtaaatccgtgggcaagtgtaaccgttttctctta attctgattgcaggtgctcagtttaataagagtgaggagcaatgcagttaccctccagactccag agaa c/a tttaccagctggtggtctcatgcagcagatccagaaaggttagttatctctgatgcgat tttgcaggtatctgactactggttccaaaattcttcttgggtttatttttgcgaattctctctct TAAAGCCACAGTGTACACTGG

FIG. 5KKKKK

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')
PAI2d20	PAI2	G	C	A	A	cds	GE318	CCACCTGATAG CCATCTGTTTT	CCAGTCTGACA CTGTGGCTTTA
PAI2d21	PAI2	T	C	-	-	noncoding	GE397	TCTGTAAAGTT CTATATCACCC ACA	GAAGTTGTTC A GAAGAGCAGAA AT
PAI2d22	PAI2	C	T	-	-	noncoding	GE397	TCTGTAAAGTT CTATATCACCC ACA	GAAGTTGTTC A GAAGAGCAGAA AT
PAI2d23	PAI2	T	A	-	-	noncoding	GE367	CAGAGTACC A CTGTAAAGCAT GT	TTACCACACCA TTTGTAAGCAG TAT
PAI2d24	PAI2	A	T	-	-	noncoding	GE367	CAGAGTACC A CTGTAAAGCAT GT	TTACCACACCA TTTGTAAGCAG TAT

FIG. 51111111

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
PAI2u1	PAI2	A	G	N	D	cds	GE315	AAACTATTAC CATGCTTAAG AACTA	TTGAATTGTAG AGAACTGCTTT GAA	AAACTATTACCATGGCTTAAGAACTATcttggttctagtagttctgtgtgtatataataaagaattcc tcttcttcttcaaggacaagctgcagataaaatccatccatctctccgtctctcagctctgc aatcaatgcataccacagg[<i>a/g</i>]attatttactggaaagtgcaataagctgttttgggtgagaag tctgcgagcttcgggaagtaagtgaaacctgtaattgaaatggctggatcccaacaagaataatg aagtcactTTCAAGCAGTCTCTACAAATTCAA
PAI2u1 0	PAI2	C	G	V	V	cds	GE333	TGGCTACTCAG AACATTTCAGTA AGT	GAGCGGTCTCT AAATTTCTGTC TAT	TGGCTACTCAGAACATTTCAGTAAGTAatttccacggttcttgattatgaacccataataatgtta tggttctgttaggcaaaatccccaaacttgctacactgaagggtctctgtagatgggataccaggatg gt[c/g]ctgggaatgctgtctactcaagtaagtggaactccatttgagaagaaactaa atgggcttatacttccgtgtaaactcggatgagacacaacaaatacatcttctcagcatatat ttcagggtctttagcaagatATAGACAGAAATTAGAGACCGCTC
PAI2u1 1	PAI2	G	A	-	-	noncoding	GE333	TGGCTACTCAG AACATTTCAGTA AGT	GAGCGGTCTCT AAATTTCTGTC TAT	TGGCTACTCAGAACATTTCAGTAAGTAatttccacggttcttgattatgaacccataataatgtta tggttct[<i>g/a</i>]taggcaaaatccccaaacttgctacctgaagggtctctgtagatgggataccag gatgtctctgggaatgctgtctactcaagtaagtggaactccatttgagaagaaactaa atgggcttatacttccgtgtaaactcggatgagacacaacaaatacatcttctcagcatatat ttcagggtctttagcaagatATAGACAGAAATTAGAGACCGCTC
PAI2u1 2	PAI2	G	A	R	H	cds	GE367	CAGAGTACCCA CTGTAAAGCAT GT	TTACCACACCA TTTGTAAGGAG TAT	CAGAGTACCCACTGTAAAGCATGTacaaaataattatttctgtgattkaaaaaaacacattnnn nnnnnnnnnnnnnnnnnnnnnnnnnnatcttaaatcgactccatatttaccctttaaataat attaggctcagc[<i>g/a</i>]cacacctgtacagatgatctgcgtgaaagctaaacattggata catagaagacctaaggctcagatctcagaaactcccatctgctggagatgttagcatgttcttgt tgctccagatgaaatgcggatgtgtccactggttgagctggtagacatcatatttaa gttctcgggctatactcactcttctgtgagatgagatgaatatATACTCTTACAAATGGTGTGG TAA
PAI2u1 3	PAI2	T	A	D	E	cds	GE315	AAACTATTAC CATGCTTAAG AACTA	TTGAATTGTAG AGAACTGCTTT GAA	AAACTATTACCATGGCTTAAGAACTATcttggttctagtagttctgtgtatataataaagaattcc tcttcttcttcaaggacaagctgcagataaaatccatccatctctccgtctctcagct ctgcaaatcaatgccacacaggaaattatttactggaaagtgcaataagctgttttgggtgagaag tctgcgagcttcgggaagtaagtgaaacctgtaattgaaatggctggatcccaacaagaataatg aagtcactTTCAAGCAGTCTCTACAAATTCAA
PAI2u1 4	PAI2	G	A	K	K	cds	GE312	GGGAAGACCAT AATTCACCAAT	CTAAGTTTCATG GATGGAAAAG ATA	GGGAAGACCATTAATTCACCAATtatgccatggcttggtttggatgtattttatgtagccttgttaa tcttcttggcttaaggaataataatcgcactctgcagaaatattactctcagacccccaggcag tagactctctagaatgtgcagaagagctagaaaaa[<i>g/a</i>]attaatctctgggtcaagactca aaccaaaaggtaaatccaagaaataatttatttacttcttccagttgagaaactctgactTATC TTTTTCCATCCATGAACCTTAG
PAI2u2	PAI2	A	G	L	L	cds	GE315	AAACTATTAC CATGCTTAAG AACTA	TTGAATTGTAG AGAACTGCTTT GAA	AAACTATTACCATGGCTTAAGAACTATcttggttctagtagttctgtgtatataataaagaattcc tcttcttcttcaaggacaagctgcagataaaatccatccatctctccgtctctcagctctgc aatcaatgcataccacaggaaattattt[<i>a/g</i>]ctggaaagtgcaataagctgttttgggtgagaag tctgcgagcttcgggaagtaagtgaaacctgtaattgaaatggctggatcccaacaagaataatg aagtcactTTCAAGCAGTCTCTACAAATTCAA

FIG. 5MMMMM

[illegible]

FIG. 5NNNNNN

[illegible]

FIG. 5000000

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')
PC1u5	PCI	C	G	Q	E	cds	GE1236	TAGGAGCAGCC CACACA	CACCTGTGCAG ACAGGA
PC1u6	PCI	G	C	S	T	cds	GE1236	TAGGAGCAGCC CACACA	CACCTGTGCAG ACAGGA
PC1u7	PCI	T	G	C	G	cds	GE1233	TTTAGGCCCT GAAAGCTA	GAAGTAAGTGT GACATGAAGGT
PC1u8	PCI	T	G	I	S	cds	GE1212	CTTCACTGACC GCTCGCC	ACCTGACCCAA AAGTCAT
PC1u9	PCI	G	A	L	L	cds	GE1236	TAGGAGCAGCC CACACA	CACCTGTGCAG ACAGGA
PC1d11	PCI	T	G	-	-	noncoding	GE334	TATGAATCCAA GGGTGAGG	GGATGAATG GGGTAGAGTAG A

FIG. 5P P P P P

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
PCId12	PCI	A	G	-	-	noncoding	GE334	TATGAATCCAA GGGTGAGG	GGAATGAATG GGGTAGACTAG A	TATGAAATCCAGGGTGAAGGctcagtggtgccaatgccccagaaacagctcaagaagctccttttc cctttccaggcagctcagctttaccttcccaaatctccatgagggtcctcctatcagctggaga aagtcctcccgagctcgggagtcagtaacgtcttcacctccctcctgactgctgtctcccgcatcagc aaccaactcaaatatccagggtgctctgagggtgggttcagaaagctcctatgcactgcttccccaaag(a /g)ctctatctctgtctctatctctctctatTCTACTCTACCCCATTTTCATTC	310
PCId13	PCI	T	A	-	-	noncoding	GE375	GAAGCTTTGCC ATTTCCTATG	GTGTGGGTGGA AGCAATCA	GAAGCTTTGCCATTTGCTATGatgacttcacctgccccctgggtggcctgggtgctgctc ccctgcagatggtgcacaaagctggtggaggtggacagctcgggaaccagagcagcgagcc acggggacaatcttcacttccaggtcgccccgctgaactctcagaggtctagtgttcaacaggcc cttctgagtgtcattgtggataaacaacatctctctctggcaaaagtgaacggccctggagt gggtctctctgaaatctacaggctcaggggtggagatgaagggggctat(a)gctatggccc atctgtagctggtagctagtgtttacgacaggttttagtgactagatgaggcattacaaataa tattactctatggatGATTGCTTCCACCCACAC	423
PCId14	PCI	T	G	V	G	cds	GE375	GAAGCTTTGCC ATTTCCTATG	GTGTGGGTGGA AGCAATCA	GAAGCTTTGCCATTTGCTATGatgacttcacctgccccctgggtggcctgggtgctgctc ccctgcagatggtgcacaaagctggtggaggtggacagctcgggaaccagagcagcgagcc acggggacaatcttcacttccaggtcgccccgctgaactctcagaggtctagtgttcaacaa ggcccttctgagtgtcattgtggataaacaacatctctctctggcaaaagtgaacggccctga gggtgggcttctctgaaatctacaggctcaggggtggagatgaagggggctatgctatggccc atctgtagctggtagctagtgtttacgacaggttttagtgactagatgaggcattacaaataa tattactctatggatGATTGCTTCCACCCACAC	423
PCId15	PCI	G	A	-	-	noncoding	GE375	GAAGCTTTGCC ATTTCCTATG	GTGTGGGTGGA AGCAATCA	GAAGCTTTGCCATTTGCTATGatgacttcacctgccccct(g/a)gtggcctgggtgctgctc tctccctgcagatggtgcacaaagctggtggaggtggacagctcgggaaccagagcagcgcc agccacggggacaatcttcacttccaggtcgccccgctgaactctcagaggtctagtgttcaaca ggcccttctgagtgtcattgtggataaacaacatctctctctggcaaaagtgaacggccctga gggtgggcttctctgaaatctacaggctcaggggtggagatgaagggggctatgctatggccc atctgtagctggtagctagtgtttacgacaggttttagtgactagatgaggcattacaaataa tattactctatggatGATTGCTTCCACCCACAC	423
PCId16	PCI	C	T	-	-	noncoding	GE375	GAAGCTTTGCC ATTTCCTATG	GTGTGGGTGGA AGCAATCA	GAAGCTTTGCCATTTGCTATGatgacttcacctgccccctgggtggcctgggtgctgctc ccctgcagatggtgcacaaagctggtggaggtggacagctcgggaaccagagcagcgagcc acggggacaatcttcacttccaggtcgccccgctgaactctcagaggtctagtgttcaacaggcc cttctgagtgtcattgtggataaacaacatctctctctggcaaaagtgaacggccctggagt gggtctctctgaaatctacaggctcaggggtggagatgaagggggctatgctatggcccattc gtatgctggtagctagtgtttacgacaggttttagtgactagatgagg(c/t)attacaaataa tattactctatggatGATTGCTTCCACCCACAC	423

FIG. 5QQQQQQ

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
PCI17	PCI	C	T	N	N	cds	GE413	GGACATCTCTG GAAAGTCAGC	TGAGGGAATTG GGTATTCTTTA GAT	GGACATCTCTGGAAAGTCAGCaccctggaccagctccaccctctctgtgagacacaccttctccct ttcagaacaaagaaacagaccaccatgcagctcttctctctctgtgctggtgcttctcagccctc agggggctcccttcaccgcccacaccccgaggagatgaagaagagatcgaggacctccatgta gggtccacgggtggccccagcagcagaaggactttacctttgacctctacaggccctggcttc cgctgccccagccagaa [c/t]atctctctctctctctctgtgagatctccatgagcctggccatg ctctccctgggggtgggtccagcacaagaatgcagatccctgggggctgggctcaacctcca gaaaagctcagagaaggagctgcacagaggctttcagcagctcttcaggaaactcaaccagccca gagatggctccagctcagctcgagctcggaatgccctttcccgacctgggtgtagacctgaggac acctcgtaagtgcctgaagacgtctgacctggcagacacttccccaccacctttagggactc tgagggggacctgaagcagatcaatgattatgtggcaagcaaacgaagggaagatctggact tgcttaagaacctcgatagcaatgcggctgtagatcatggtagaattacatctctcttaaggtaag gcccttggggcccaaacctgcacttctctggctttctgtgctgttttTCTAAAGAATACCCAAT TCCTCA
PCI18	PCI	C	T	P	P	cds	GE413	GGACATCTCTG GAAAGTCAGC	TGAGGGAATTG GGTATTCTTTA GAT	GGACATCTCTGGAAAGTCAGCaccctggaccagctccaccctctctgtgagacacaccttctccct ttcagaacaaagaaacagaccaccatgcagctcttctctctctgtgctggtgcttctcagccctc agggggctcccttcaccgcccacaccccgaggagatgaagaagagatcgaggacctccatgta gggtccacgggtggccccagcagcagaaggactttacctttgacctctacaggccctggcttc cgctgccccagccagaaacttctctctctgtgagatctccatgagcctggccatgctct ccctgggggtgggtccagcacaagaatgcagatccctggagggtgggctcaacctccagaaa agctcagagaaggagctgcacagaggcttcagcagctcttcaggaaactcaaccagccagaga tggtctccagctgagctcggaatgccctttcccgacctgggtgtagacctgaggacacct tcgttaagtgcctgaagacgtctgacctggcagacacttccc [c/t]accaactttagggactc tgagggggacctgaagcagatcaatgattatgtggcaagcaaacgaagggaagatctggact tgcttaagaacctcgatagcaatgcggctgtagatcatggtagaattacatctctcttaaggtaag gcccttggggcccaaacctgcacttctctggctttctgtgctgttttTCTAAAGAATACCCAAT TCCTCA
PCI19	PCI	T	C	F	F	cds	GE413	GGACATCTCTG GAAAGTCAGC	TGAGGGAATTG GGTATTCTTTA GAT	GGACATCTCTGGAAAGTCAGCaccctggaccagctccaccctctctgtgagacacaccttctccct ttcagaacaaagaaacagaccaccatgcagctcttctctctctgtgctggtgcttctcagccctc agggggctcccttcaccgcccacaccccgaggagatgaagaagagatcgaggacctccatgta gggtccacgggtggccccagcagcagaaggactttaccttt [t/c]gacctctacaggcccttgg cttccgtgccccagccagaaacttctctctctgtgagatctccatgagcctggccatg ctctccctgggggtgggtccagcacaagaatgcagatccctggagggtgggctcaacctcca gaaaagctcagagaaggagctgcacagaggcttcagcagctcttcaggaaactcaaccagccca gagatggctccagctcagctcggaatgccctttcccgacctgggtgtagacctgaggac acctcgtaagtgcctgaagacgtctgacctggcagacacttccccaccacctttagggactc tgagggggacctgaagcagatcaatgattatgtggcaagcaaacgaagggaagatctggact tgcttaagaacctcgatagcaatgcggctgtagatcatggtagaattacatctctcttaaggtaag gcccttggggcccaaacctgcacttctctggctttctgtgctgttttTCTAAAGAATACCCAAT TCCTCA

FIG. 5RRRRRR

[illegible]

FIG. 5SSSSS

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
PCIu5	PCI	C	T	A	V	cds	GE413	GGACATCTCTG GAAAGTCAGC	TGAGGGAATTG GGTATTCTTTA GAT	GGACATCTCTGAAAGTCAGCacctggaccagctccacccctctctgaggacaccttcttcccttctcagaacaaagacagccaccatgcagctcttctctctctgtgcttggtcttctcagccctcaggggccctcccttcaccgcccacacccccggagatgaagaagagtcgaggacccctccatgtagtgcacaggtggccccagcagcagaaggaacttacccttgacctctacagggc/c/t)cttggcttcgctgccccagcagacacatcttcttctccctgtgagcatctccatgagcctggccatgctctccctggggctgggtccagcacaagatccagcagctctcagcagctctcaggaactcaaccccca gaaagctcagagaagagctgcacagaaggttccagcagctctcaggaactcaaccccca gagaaggctccagctgagcctcgcaatgccccttccagcagctgggtggtgagcctgcaggacacctcgtaagtccatgaagagctgtacctggcagacacttccccaccaactttagggactctgcaggggccatgaagcagatcaatgattatgtggcacaagcaaacgagggcaagattgtggaacttgcctaaagacctcgatagcaatgcgtcgatcatggtgaattacatcttctttaaaggtaag gcccctggggcccaaacctgcacttcttggcttctctgctgttcttATCTAAAGATACCCCAAT TCCCTCA
PCIu6	PCI	A	G	K	E	cds	GE413	GGACATCTCTG GAAAGTCAGC	TGAGGGAATTG GGTATTCTTTA GAT	GGACATCTCTGAAAGTCAGCacctggaccagctccacccctctctgaggacaccttcttcccttctcagaacaaagacagccaccatgcagctcttctctctctgtgcttggtcttctcagccctcaggggccctcccttcaccgcccacacccccggagatgaagaagagtcgaggacccctccatgtagtgcacaggtggccccagcagcagaaggaacttacccttgacctctacagggccttggcttc cgtgccccccagcagacacatcttcttctccctgtgagcatctccatgagcctggccatgctct cctggggctgggtccagcacaagatgcagatcctggaggcctggcctcaacccctccagaa agctcagag/a/g)aggagctgcacagaaggttccagcagctctcaggaactcaaccccca gagaaggctccagctgagcctcgcaatgccccttccagcagctgggtggtgagcctgcaggacacctcgtaagtccatgaagacgctgtacctggcagacacttccccaccaactttagggacttgcaggggccatgaagcagatcaatgattatgtggcacaagcaaacgagggcaagattgtggaacttgcctaaagacctcgatagcaatgcgtcgatcatggtgaattacatcttctttaaaggtaag gcccctggggcccaaacctgcacttcttggcttctctgctgttcttATCTAAAGATACCCCAAT TCCCTCA
PCIu7	PCI	T	A	F	I	cds	GE413	GGACATCTCTG GAAAGTCAGC	TGAGGGAATTG GGTATTCTTTA GAT	GGACATCTCTGAAAGTCAGCacctggaccagctccacccctctctgaggacaccttcttcccttctcagaacaaagacagccaccatgcagctcttctctctctgtgcttggtcttctcagccctcaggggccctcccttcaccgcccacacccccggagatgaagaagagtcgaggacccctccatgtagtgcacaggtggccccagcagcagaaggaacttacccttgacctctacagggccttggcttc cgtgccccccagcagacacatcttcttctccctgtgagcatctccatgagcctggccatgctct cctggggctgggtccagcacaagatgcagatcctggaggcctggcctcaacccctccagaa agctcagagaagagctgcacagaaggttccagcagctctcaggaactcaacccccaagaga tggctccagctgagcctcgcaatgccccttccagcagctgggtggtgagcctgcaggacaccttcgtaagtccatgaagacgctgtacctggcagacacttccccaccaactttagggactctgca gggcccatgaagcagatcaatgattatgtggcacaagcaaacgagggcaagattgtggaacttgcctaaagacctcgatagcaatgcgtcgatcatggtgaattacatcttctttaaaggtaag gcccctggggcccaaacctgcacttcttggcttctctgctgttcttATCTAAAGATACCCCAAT TCCCTCA

FIG. 5TTTTTT

FIG. 5UUUUU

[illegible]

FIG. 5VVVVV

[illegible]

FIG. 5 WWW

[illegible]

FIG. 5XXXXXX

[illegible]

FIG. 5YYYYYY

[illegible]

FIG. 5ZZZZZ

[illegible]

FIG. 5A

[illegible]

FIG. 5BBBBBBB

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
SELPd2 4	SELP	C	T	P	L	cds	GE954	TGCCACCCCT GAAGAT	GGGCTGGGGCT GTCCAT	TGCCACCCCTGAAGATtctctgaacaggaacatgatctgccttcattctgcaaaagcattcca gcatcagctcagctcagcttcagttgtgaagagggatttgcattagttggac c/t ggaagtgt gtcgaatgcacagcctcgggggtATGGACAGCCCCAGCCCC	170
SELPd2 5	SELP	G	A	V	M	cds	GE446	CTCTGCAGTG AGAGAGTG	CACTGTGGAGG CTTATTG	CTCTGCAGTGAGAGAGTGtggagaacttgagctccctccctcaacac g/a tgctcatgaactgcag ccacctctggaaactctctttaaactcgcagtgagcttccactgcactgaactgaagggtaccag taaatgggcccacagcaagctgggaatgcttggtctctggaaactctggacAAATAAGCCTCCACAGTG	194
SELPu1	SELP	A	C	N	T	cds	GE462	TACTCTAGCCA TCAAGTGCC	ATATTATTACC TTTCAGGTTG	TACTCTAGCCATCAAGTGCCcagaactcttggccccagagagggcagcctggattgttctgaca ctctgtggagaattcaaatgttggctccacctgtcatctctctgttaaca a/c tggtctttaaagct ggagggcccacaataatgtggaatgcacaactcttggaagatggctcagctactccacCAACCTGCA AAGGTAATAATAT	208
SELPu1 0	SELP	A	G	T	A	cds	GE466	TCTCTCAGCT GTGCAGT	AAATCCTACC CTCACAGG	TCTCTCAGCTGTGCAGTgtcagcaccttggaaagccccccagtgaaagga a/g ccatggagctgtgt tcatcgcctcactgcttcttgcctatggctccacctgtcatctctctgttaacaatggctttaaagct tgagggccttgacatgctccgctgcattgactctggacactggctcagctactccacCAACCTGCT GAGGTAGGATTTT	209
SELPu1 1	SELP	T	C	T	T	cds	GE462	TACTCTAGCCA TCAAGTGCC	ATATTATTACC TTTCAGGTTG	TACTCTAGCCATCAAGTGCCcagaactcttggccccagagagggcagcctggattgttctgaca c t/c ctgtggagaattcaaatgttggctccacctgtcatctctctgttaacaatggctttaaagct ggagggcccacaataatgtggaatgcacaactcttggaagatggctcagctactccacCAACCTGCA AAGGTAATAATAT	208
SELPu1 2	SELP	A	G	N	D	cds	GE462	TACTCTAGCCA TCAAGTGCC	ATATTATTACC TTTCAGGTTG	TACTCTAGCCATCAAGTGCCcagaactcttggccccagagagggcagcctggattgttctgaca ctctgtggagaattcaaatgttggctccacctgtcatctctctgt a/g acaatggctttaaagct ggagggcccacaataatgtggaatgcacaactcttggaagatggctcagctactccacCAACCTGCA AAGGTAATAATAT	208
SELPu1 3	SELP	A	G	T	T	cds	GE421	TTTGTAGCAGG ACCATTGA	TCCTATTTACC TTTTGTCTGA	TTTGTAGCAGGACCATTGACTatccaggaaagccctgacttactttgggtggaggggtggcttctac a/g atagggtctgataatgggtggagcgtctctctggctttgctaagaagcgtttTCAGACAAAAA GGTAAATAGGA	141
SELPu1 4	SELP	G	A	P	P	cds	GE483	AACCAGAAAGA AGTGCCAG	AAGGCCCTACC TGTGTAA	AACCAGAAAGAGTGCCAGGatggacttatcatlacagcaaaaaagcactactcatggaatatttc ccgtaaataactgccagaatcgctacagacttagtgggccatccagataaaaaatgaaattgatt acctcaataaggctcctaccctactacagctcctactactgtggtatccgaaagaacaaataag acatggacatgggtgggaaccaaaggctctcccaacgggctgagaactgggctgataatga acctaaacaaaggaacacgaggtgcgtggagatacatacaaggtcc g/a tcagcc cctggcaagtggaaatgatgagcactgcttgaaagaaaaagcagcacttggtTACACAGGTAGGGC CTT	393
SELPu1 5	SELP	T	C	F	S	cds	GE466	TCCTCAGCT GTGCAGT	AAATCCTACC CTCACAGG	TCCTCAGCTGTGCAGTgtcagcaccttggaaagccccccagtgaaaggaacatggactgtgttcat ccgctcactgctt t/c tgctatggctccagctgcaaat tgagtgccagcccggtctacagag tgagggccttgacatgctccgctgcattgactctggacactggctgcaccttgccaaCCTGT GAGGTAGGATTTT	209
SELPu1 6	SELP	C	T	S	F	cds	GE445	ATCCTTAGCT TTGCAGTG	ACACTCTTACC TTGCATTTC	ATCCTTAGCTTTGCAGTgcccaggatctccccagttccaaataggggcccggtgaactgctcccac cctctgggtgctcttaggtacagctcagctgcagcttccacttcaatgaaggcttgcctcctgggt ggaggaagtgtgctacagtgttggtcacttggaactggaaatt c/t tggtcctccaGAATGC CAAGGTAAGAGTGT	209

FIG. 5CCCCCCC

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
SELPu17	SELP	C	T	D	D	cds	GE454	TCCACTTAGCTATTTCGTGAGCGCTGGAGAGTCTCTCCAGGAGCATGGATTGCTCTCCA ATTTCTGTGTC	ACAGTACCTTG ACAGACTGGG	TCCACTTAGCTATTTCGTGAGCGCTGGAGAGTCTCTCCAGGAGCATGGATTGCTCTCCA tccttgagagcgtttcagtgatga(c/t)accaactgtagctccgctgtgctggaagggtttcagtc tgagaggagcgcgatatagttcggtgtgataaacttggacagtggaacagaccagcCCCAGTCTGT CAAGGTACTGT	206
SELPu18	SELP	A	T	S	C	cds	GE446	CTCCTGCAGTCAGAGAGTGTGGAGAACTTGGAGTCCCTCAACAGTGTCTCATGAAGTGCAGCCAC AGAGAGTG	CACTGTGGAGG CTTATTATTG	CTCCTGCAGTCAGAGAGTGTGGAGAACTTGGAGTCCCTCAACAGTGTCTCATGAAGTGCAGCCAC cctctgggaaacttctctttaaactgcagtcagctccactgcactgacgggtaccaggtaaa tggggccc(a/t)lgcaagctgggaatgcttggcttctggaaacttgggaCAATAAGCCTCCACAGTG	194
SELPu19	SELP	G	A	S	N	cds	GE466	TCCTCTCCAGCTGTGTCAGTGTGAGCACTTGGAGTCCCTCAACAGTGTCTCATGAAGTGCAGCCAC GTCCAGT	AAATCTCTACC CTCACAGG	TCCTCTCCAGCTGTGTCAGTGTGAGCACTTGGAGTCCCTCAACAGTGTCTCATGAAGTGCAGCCAC tcatccgctcactgcttttgcctatggctcagctgcaaatgtgagtgccagcccggtctacagag tgaggggcttggacatgctcgcgtgcatgactctgyacactggctgacaccttgcuaaCCCTGT GAGGTAGGATTT	209
SELPu20	SELP	T	C	N	N	cds	GE462	TACTCTAGCCATCAAGTGCAGAACTCTTGGCCCAAGAGCGGCGAGCCTGGATTGTTCTTGACA TCAAGTGCC	ATATTATTACC TTTGCAGGTTG	TACTCTAGCCATCAAGTGCAGAACTCTTGGCCCAAGAGCGGCGAGCCTGGATTGTTCTTGACA ctcgtggagaattcaatgttggctccacctgtcatttctctgttaacaatggctttaagctggag ggggccaa(t/a)aatgtggaatgcacaacttctggaagatggtcagctactccacCAACCTGCA AAGGTAATAATAT	208
SELPu20	SELP	C	G	S	*	cds	GE483	AACCAGAAAGA AGTGGCAG	AAGGCCCTACC TGTGTAA	AACCAGAAAGAAGTGGCAGCATGGACTTATCATTACAGCACAAAAGCATACTTC/g)atggaata ttcccgtaaataactgcccagaatcgctacacagactttagtgccactccagaataaaaaatgaaatt gattacctcaataaggctcctactacactcctactactggtatgggattccgaaagaacaa taagacatggacatgggtgggaacaaagggctctcaccacagaggtgagaaactgggctgata atgaacctaaacaaaggaacaaacgagggactgctggaagatatatacatcaagagtcctgcagcc cctggcaagtgggaatgatgagcactgcttgaagaaaaagcagcattgtgTTACACAGT'AGGGCC CTT	393
SELPu21	SELP	T	A	N	K	cds	GE483	AACCAGAAAGA AGTGGCAG	AAGGCCCTACC TGTGTAA	AACCAGAAAGAAGTGGCAGCATGGACTTATCATTACAGCACAAAAGCATACTTC/g)atggaata ccgtaaataactgcccagaatcgctacacagactttagtgccactccagaataaaaaatgaaatt acctcaataaggctcctactacactcctactactggtatgggattccgaaagaacaa(t/a)aagacatggacatgggtgggaacaaagggctctcaccacagaggtgagaaactgggctgata atgaacctaaacaaaggaacaaacgagggactgctggaagatatatacatcaagagtcctgcagcc cctggcaagtgggaatgatgagcactgcttgaagaaaaagcagcattgtgTTACACAGT'AGGGCC CTT	393
SELPu22	SELP	T	A	C	S	cds	GE451	TCACAACAGGC ATAGCAT	CCCTCTGCATGC TGGAGTT	TCACAACAGGCATAGCATCacttctactccaggggttgcaatgtccagccctcaccactcctggg cagggaaacctgactgtaggcatcatccgggaaccttgggtttaaataccacttgttactttgg ctgcaacgctggttcaactcataggagacagcactctcagc(t/a)gcagaccttcaggacaa tggacagcagcTAACCTCCAGCATGCAGAGG	224
SELPu3	SELP	T	A	N	K	cds	GE462	TACTCTAGCCATCAAGTGCAGAACTCTTGGCCCAAGAGCGGCGAGCCTGGATTGTTCTTGACA TCAAGTGCC	ATATTATTACC TTTGCAGGTTG	TACTCTAGCCATCAAGTGCAGAACTCTTGGCCCAAGAGCGGCGAGCCTGGATTGTTCTTGACA ctcgtggagaattcaatgttggctccacctgtcatttctctgttaacaatggctttaagctggag ggggccaa(t/a)aatgtggaatgcacaacttctggaagatggtcagctactccacCAACCTGCA AAGGTAATAATAT	208
SELPu4	SELP	T	G	S	A	cds	GE462	TACTCTAGCCATCAAGTGCAGAACTCTTGGCCCAAGAGCGGCGAGCCTGGATTGTTCTTGACA TCAAGTGCC	ATATTATTACC TTTGCAGGTTG	TACTCTAGCCATCAAGTGCAGAACTCTTGGCCCAAGAGCGGCGAGCCTGGATTGTTCTTGACA ctcgtggagaattcaatgttggctccacctgtcatttctctgttaacaatggctttaagctggag ggggccaaatgtggaaatgcacaact(t/g)ctggaagatggtcagctactccacCAACCTGCA AAGGTAATAATAT	208

FIG. 5DDDDDDDD

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
SELPu5	SELP	T	G	L	V	cds	GE451	TCACAAACAGGC ATAGCAT	CCCTCTGCATGC TGGAGTT	TCACAACAGGCATAGCATcacttccctactccagggtctg]tgcaatgtccagccctcaccactcc tggcagggaaccatgtactgtaggcatcatccgggaacctttgggttttaataccacttggctact ttggctgcaacgctggattcacactcatagagacagcactctcagctgcagaccttcaggacaa tggacagcagcAACTCCAGCATGCAGAGG	224
SELPu6	SELP	A	G	M	V	cds	GE466	TCTCTCAGCT GTGCAGT	AAAATCCTACC CTCACAGG	TCTCTCCAGCTCTGCAGTgtcagcaccctggaagcccccagtggaagaaacctggactgtgttcac cggctcactgtcttggctcctagctcagctgcaaatttgagtgccagccggctacagagtgag gggcttggac]a/g]tgctccgctgacttgactctggacactggctgcacccttgccaaCTGTGT GAGGTAGGATTTT	209
SELPu7	SELP	T	C	C	C	cds	GE462	TACTCTAGCCCA TCAAGTGCC	ATATTATTACC TTTGCAGGTG	TACTCTAGCCCATCAAGTGCCcagaactcttggccccagagcaggggcagcctggattgttctgaca ctcgtggagaattcaatgttggctccacctg]t/c]catttctctgtacaatggcttttaagct ggaggggcccaataatgtgggaatgcacaacttctggaagatggctcactccacCAACTTGCA AAGGTAAATAAT	208
SELPu8	SELP	A	C	T	P	cds	GE452	CAGCTGTGAAA TGCTCAGA	AAAATTGTACC TTGGCAGG	CAGCTGTGAAATGCTCAGAActacatgttaataagcccaatagcgatgaactgctcccaacctctgg ggaaacttcagttatggatcaatctgctcttccacttctagagggccagttacttaattggctc tgcaaaaacagcatgccaaagaaatggccactgggtca]a/c]ctaccgtgccaaCTGCCAAGGT ACAATTTT	203
SELPu9	SELP	A	T	K	*	cds	GE483	AACCAGAAAGA AGTGCCAG	AAGGCCCTACC TGTGTNA	AACCAGAAAGAAGTGCCAGcatggacttatcatcacagacaaaagacatactcatggaatatttc ccgt]a/t]aatactgccagaatcgtcacacagactttagtgccatccagaaataaaatgaaatt gattacctcaataaggtcctaccctactacagctcctactagattggatgccgaagaaacaa taagacatggacatgggtgggaaacaaaaggctctcacaacaggctgagaaactgggctgata atgaacctacaacaaaaggaaacagcaggactcgtggagatatacatcaagagtcctgcagcc cctggcaggtggaaatgatgagcactgtctgaagaaaagcagcgttctgtTACACAGGTAGGGC CTT	393
SHBGd3	SHBG	T	C	-	-	noncoding	GE519	TGACATGTCCC TACTCAGCTTT	AACCAGGGGAC TGGGTC	TGACATGTCCCTACTCAGCTTTgttgg]t/c]tctctcttctgatagagtggccacgacctcc ggctgtccacctcagcaatggccaggaacagacctatcgctgtcatgaccttggacctcacca agatcacaaaglatggggttggcctagccttGACCCAGTCCCCTGGTT	179
SHBGd4	SHBG	C	T	P	L	cds	GE603	GGCAGAAACAG ATCCAGG	CCAGCAGTGCT TTACCNAGTC	GGCAGAAACAGATCCCAGggcctctgatttggcttcccaccttctgcagggtggaggtcaagat ggagggggactctgtgtgtggggtggatggggaggaggtgtgtgcgctggagacaggtctctg ggcccttgaccagcaaacgcatccatcatgaggtatggcttgggggggtgctcttcccgt tccaaacctcgggtgc]t/l]ggtaactacacccccaggggtggaaaccttagccaaGACTTGGTAA AGCACTGCTGG	271
SHBGd5	SHBG	G	A	D	N	cds	GE605	CACCTTAATGC TCTAATGCCA	TTGCTAGTGCA CAGTGAATATT TG	CACCTTAATGCTCTAATGCCACcttggcactacctccctcttagggaa]g/alactctccacct cttttgcctgaaatggccttgggcacaaaggtcagaggtggatggagccaggccctggacaga agcatgagatctggactcagctgccccagagccagggcaatggcactgagcttcccat aagctccacctaaagaaccccttggaaagtactgatttcatattcaacAAATATTCACT GTGCACCTAGCAA	272
SHBGd6	SHBG	C	A	T	N	cds	GE605	CACCTTAATGC TCTAATGCCA	TTGCTAGTGCA CAGTGAATATT TG	CACCTTAATGCTCTAATGCCACcttggcactacctccctcttagggaaagactcttccacctctt ctgctgaaatggccttgggcacaaaggtcagaggtggatggagccaggccctgaacagaagcc atgagatctgg]c/a]tcacagctgccccagagccagggcaatggcactgagcttcccat aagctccacctaaagaaccccttggaaagtactgatttcatattcaacAAATATTCACT GTGCACCTAGCAA	272

FIG. 5EEEEEE

[illegible]

FIG. 5FFFFF

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
SLC6A1 u5	SLC6A1	C	A	I	I	cds	GE1097	AGGAGGAGGG TCACCACT	CCTGGTCTTACA GTGAGGGTCA	AGGAGGAGGGTACACAGTcaccactttctccagggtggctacttttccagccacataccctt acat(c/a)atngctgnatcatcctngttcttccgtggagtgacgtgccgggccaaggagggg cactcttctacatcacaccaacttcgcgaagctgtctgactccgaggtgagtgccctccca gcccggattctgagccaccactagcagcttgagataagcttttgcatctttnTGACCTTCACT GTAGACCAGG	270
SLC6A1 u6	SLC6A1	G	T	T	T	cds	GE1078	CCGGCTCCAC CAGCTC	CCTGTTCTTTT ATCCAGTGCC	CCGGCTCCACACAGCTCgtgaactttctctccctccactctglttgaccaggcgcaacatgcac cagatgacggagcgggtggaagcaggtcagatccgctggccactggccatcac(g/t)ctgg ccacgctggatcctgtgtatctctgctggaagggtgttgctggactgggaaggtaagg gatatagtgcacagtggggacaggagGCACTCGATTAAGGACAGG	243
SLC6A1 u7	SLC6A1	T	A	I	N	cds	GE1044	AATTCGGTATT AAATGGACG	GTGGTTTGTGT TGGATGGGG	AATTCGGTATTAAATGGACGngggtntgttctccgtccaggcgtgggcttgcggctgctgtg ctatcattctgtgctgaacatctactaca(lt/a)cgctacatctctctggccatttactacctgt acaactcttcacacgggtgagtggtggtcactttgaccccCCCATCCAAACAAACCCAC	188
SLC6A1 u8	SLC6A1	G	C	C	S	cds	GE1250	GCAGACACTGC CGTGGA	CTCCTGCCGGG CTCTGG	GCAGACACTGCCTGGAaacagtgcgacaacccctgggaacacagaccgt(g/c)cttctccaac tacagcatggttcacactacacacatgaccagcgtgtggtggagttctcgggagtgatggca taatcaggggatggcagggaagggtcagCCAGAGCCCGCAGGAG	180
SLC6A3 d13	SLC6A3	G	A	R	Q	cds	GE1045	TGCAGACGTGG CGTGCT	CCCATGTCTAC AGGCCCAAT	TGCAGACGTGGCTGCTgcacctccacacagagccatggcatcgacacctgggctccgc(g/a) lgtggcagctcacagcctgctggtggtcgtcgtgctgctctactcagcctctggaaagggc gtgaagacctcagggaagggtgaggtcgggggtcaccatTTGGGCTGTAGACATGGG	188
SLC6A3 d14	SLC6A3	G	A	V	M	cds	GE1128	TGAAGACCAAG AGGGAAG	CTTACCGCCAC CATTTT	TGAAGACCAAGAGGGAAGaagcacagaaatctctcaactccagtggtgccatgagtaagagcaaa tgcctcgtgggactcatgtcttcggtggtggcccggtcgaagagccaatgccggggccgaa ggaggtggagctcatcttgcaggagcagaaacggagtgcagctcacagctccacccctacca acccgcgagagcccc(g/a)lgtggagccacaggtcgggagacctggggaagagatcgactt tctcctgctcgtcatgtggttctgtgtggacctggcacaagctctggcggttccccctactgtgt acaAAATGGTGGCGGTAAAG	345
SLC6A3 u1	SLC6A3	C	G	L	V	cds	GE1045	TGCAGACGTGG CGTGCT	CCCATGTCTAC AGGCCCAAT	TGCAGACGTGGCTGCTgcacctccacacagagccatggcatcgacacctgggctccgcgggtg gcag(c/g)tcacagcctgctggtggtcgtggtcgtgctgctctactcagcctctggaaagggc gtgaagacctcagggaagggtgaggtcgggggtcaccatTTGGGCTGTAGACATGGG	188
SLC6A3 u10	SLC6A3	G	T	P	P	cds	GE1128	TGAAGACCAAG AGGGAAG	CTTACCGCCAC CATTTT	TGAAGACCAAGAGGGAAGaagcacagaaatctctcaactccagtggtgccatgagtaagagcaaa tgcctcgtgggactcatgtcttcggtggtggcccggtcgaagagccaatgccggggccgaa ggaggtggagctcatcttgcaggagcagaaacggagtgcagctcacagctccacccctacca accc(g/t)cggcagagccccgtggagggccaggatcgggagacctggggaagagatcgactt tctcctgctcgtcatgtggttctgtgtggacctggcacaagctctggcggttccccctactgtgt acaAAATGGTGGCGGTAAAG	345
SLC6A3 u11	SLC6A3	C	A	L	I	cds	GE1045	TGCAGACGTGG CGTGCT	CCCATGTCTAC AGGCCCAAT	TGCAGACGTGGCTGCTgcacctccacacagagccatggcatcgacacctgggctccgcgggtg gcagctcacagcctgctggtggtggtcgtggtcgtgctctactcagc(c/a)cttggaaagggc gtgaagacctcagggaagggtgaggtcgggggtcaccatTTGGGCTGTAGACATGGG	188
SLC6A3 u12	SLC6A3	G	C	V	V	cds	GE1045	TGCAGACGTGG CGTGCT	CCCATGTCTAC AGGCCCAAT	TGCAGACGTGGCTGCTgcacctccacacagagccatggcatcgacacctgggctccgcgggtg gcagctcacagcctgctggtggtcgtggtcgtggtcgtgctctactcagcctctggaaagggcgt /c/aagacctcagggaagggtgaggtcgggggtcaccatTTGGGCTGTAGACATGGG	188
SLC6A3 u2	SLC6A3	A	G	I	V	cds	GE1043	CAAGGGCCCTG CCTGTG	ACAGACGCTC GCAGAGC	CAAGGGCCCTGCTGTGcagcctggtgacactgttcttcttgcagggtggtatgg(la/g)tcaca gccaacctgccatacgtggctcctcactgccctgctcctggtgggtcaccctccctggagccat agcggcatcagagcatactgagcgttgacttctccgctcctcagcctcctcagcctcctcgt	187

FIG. 5GGGGGGG

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
SLC6A3 J3	SLC6A3	G	A	S	S	cds	GE990	TCAGGGCCAGGCTGAT	CACGGCGCTGT CGATAC	TCAGGGCCAGGCTGATcttcatcatctaccggaagccatcgcaacgctcctctgtctctc g/124 algcctgggcccgtggtctcttcttcatcatgctgctcaccctggGTATGACAGCGCCGCTG
SLC6A3 J4	SLC6A3	C	T	A	A	cds	GE1043	CAAGGGCCCTGCTG	ACAGAGCCCTC GCAGAGC	CAAGGGCCCTGCTGctgacagctggtgacactgtcttcttgcagggtggtatggatcacagc c/187 /c accatgccatacgtggtctcactgctcgtcgtggtggtcaccctccctggagccat agacggcatcagagcataccctgagcgttgacttctacggGCTCTGCGAGGCGCTGT
SLC6A3 J5	SLC6A3	C	T	A	A	cds	GE1273	CTTTGTCTCTGG CACCGC	AGCCTTCTCTGG TGGCCTCA	CTTTGTCTCTGGCACCGCgggctgagagctgctgacctccgtatctgctggtgagcttcgtggt cgtggtcagcatgtgaccttcagacccccacactcaggaagcctacatcttcccagactgggcca acgagctgggctgggtcactgcacacatctccatggccatggtgcccactctatgcggc c ta caagtctcgcagcctgctgggtctcttcgagaggtggttatctggaagggtggttaccctcaccgga gltGTAGGCCACCAGAAAGGCT
SLC6A3 J6	SLC6A3	C	T	N	N	cds	GE1128	TGAAGACCAAG AGGGAAG	CTTACGCCAC CATTTT	TGAAGACCAAGAGGGAAGaagcacagaaattctcctaactccagtgctgccccatgagtaagagcaaa tgctccgtgggactcatgtcttcctggtggccccggctaaaggagcccaatgcgtgggcccga ggaggtggagctcatctgtcgaaggagcaaacggagtgacgtccacagctccaccctcacc accacccggcagagccccgtggagggccccaggatcgggagacctggggcaagaagatcgactt tctcctgctcgtcatgggtctgtgtgacctggccaaagctctggcggttcccctacctgtgct acaAAATGTGTGGCGGTAAAG
SLC6A3 J7	SLC6A3	C	T	P	P	cds	GE1128	TGAAGACCAAG AGGGAAG	CTTACGCCAC CATTTT	TGAAGACCAAGAGGGAAGaagcacagaaattctcctaactccagtgctgccccatgagtaagagcaaa tgctccgtgggactcatgtcttcctggtggccccggctaaaggagcccaatgcgtgggcccga ggaggtggagctcatctgtcgaaggagcaaacggagtgacgtccacagctccaccctcacc accacccggcagagcccc c t gtggagggccccaggatcgggagacctggggcaagaagatcgactt tctcctgctcgtcatgggtctgtgtgacctggccaaagctctggcggttcccctacctgtgct acaAAATGTGTGGCGGTAAAG
SLC6A3 J8	SLC6A3	G	C	R	R	cds	GE1128	TGAAGACCAAG AGGGAAG	CTTACGCCAC CATTTT	TGAAGACCAAGAGGGAAGaagcacagaaattctcctaactccagtgctgccccatgagtaagagcaaa tgctccgtgggactcatgtcttcctggtggccccggctaaaggagcccaatgcgtgggcccga ggaggtggagctcatctgtcgaaggagcaaacggagtgacgtccacagctccaccctcacc accacccggcagagccccgtggagggccccaggatcgggagacctggggcaagaagatcgacttctc ctgtccgtcatgtgcttggctggacctggccaaagctctggcg g ctcccctacctgtgct acaAAATGTGTGGCGGTAAAG
SLC6A3 J9	SLC6A3	C	G	A	G	cds	GE1128	TGAAGACCAAG AGGGAAG	CTTACGCCAC CATTTT	TGAAGACCAAGAGGGAAGaagcacagaaattctcctaactccagtgctgccccatgagtaagagcaaa tgctccgtgggactcatgtcttcctggtggccccggctaaaggagcccaatgcgtgggcccga ggaggtggagctcatctgtcgaaggagcaaacggagtgacgtccacagctccaccctcacc accacccggcagagccccgtggagggccccaggatcgggagacctggggcaagaagatcgacttctc ctgtccgtcatgtgcttggctggacctggccaaagctctggcg g ctcccctacctgtgct acaAAATGTGTGGCGGTAAAG
SLC6A4 J10	SLC6A4	A	C	K	N	cds	GE1113	CATCTCTATCT GAGTGGATATT GT	TTTCTCCCAA AACAATTAGTA GTC	CATCTCTATCTGAGTGGATATTGTTaagggtttttaaagtgtcttggaattctgttttagttcat catcttgagtttcttgatgagccgcacacactacgactttcccaataataatctcttacttggga gtatcatcttggttactgcataggaacctcatcttctatctgcatccccacatatagattat cggttgatcatcactccagggacatttaa a/c gaggtacgtgctagttagtgatatactgtgt gtgttcAGACTACTAATTGTTTTCGGAGAAAA
SLC6A4 J9	SLC6A4	G	A	T	T	cds	GE1071	TGTGACATCTT TGTAGGACAGG T	CCTCCTTCTCT CTTCATCC	TGTGACATCTTGTAGACAGGTTtgtcaaccacactctctctctctctctctctctgtctcaggtcc cagctctctctctcatcagctatgcagaagcagatagccacatgcacagctccacttctctggcca tcatctctctcttgatgttaactcagctgggttggagagcac g/a gtgaggaatgggaagg caaatggcctctgggagcctgacagcaggaagcagc

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
SLC6A4 u1	SLC6A4	G	C	K	N	cds	GE1124	CCGTGGGCCA CCCTTA	TGGCCTGCCCC TAACAGG	CCGTTGGGCCACCCCTTAAactcggcctctgtctctccatcttaccactgcccagggtatggttat tgccatctgcacatctgctcttaccatctgcttactactaacacacatcatgctcctgggccttat actactctcatctcctctccacggaccagctgctcctggaccagtgcacaa[g/c]aactcctggaa cactggcaactgcaccaattactctccgaggacaacatcacctggaccctccattccacgtccc ctgctgaagaattttacacgcgccacgtaatgcacgtatgaggttggCCTGTTAGGGGCAGG CCA
SLC6A4 u2	SLC6A4	C	A	-	-	noncoding	GE1027	AGCATCAGTAA CCTGCACACTC	ATTTCAGGCC CGACTGGT	AGCATCAGTAACTGCACACTCTctctccctaggctctctcctggaggcgaaggcacttgccttgcctt ctattgcagaataacaagggtcttagccacagagtgg[c/a]tggaagtggaaagaagaacaa atggtgagcagcagggaagtttaaggACCAGTCGGGCTTGCATAAT
SLC6A4 u3	SLC6A4	A	C	-	-	noncoding	GE1027	AGCATCAGTAA CCTGCACACTC	ATTTCAGGCC CGACTGGT	AGCATCAGTAACTGCACACTCTctctccctaggctctctcctggaggcgaaggcacttgccttgcctt ct[a/c]ttgcagaataacaagggtcttagccacagagtgtgctggcaagtggaaagaagaacaa atggtgagcagcagggaagtttaaggACCAGTCGGGCTTGCATAAT
SLC6A4 u4	SLC6A4	G	C	G	A	cds	GE1145	AAATCCAAGCA CCCAGAGAT	ACTCCGGGTCA CAGCCCAC	AAATCCAAGCACCCAGAGATcaattgggataccttggcagatggacatcagtgctcatlactaacc agcaggatggagacgacgccccttgagattctcagaagcagctatcagctgtgagatggagaaga tctgtcaggaaaacggagttcttcagagaagttgttcccaaccagggaacaaagtggagtcgggc aaatatccaatgggtactcagcagttccaaagctcctgtgctgg[g/a]agatgacacacggcactc tatccacgcgacacacacacccctagtggtgagcttcatcaaggggaaacgggacctggggca agaaggtggatttctctctcagtgattgctatgcttggacctgggcaatgtctgctgctgctc ccctacatatgttaccagaatggagggggtcagtatcacaggctgcagtagtggctgtgaccca ggGTGGGCTGTGACCCCGAGT
SLC6A4 u5	SLC6A4	G	C	M	I	cds	GE1124	CCGTGGGCCA CCCTTA	TGGCCTGCCCC TAACAGG	CCGTTGGGCCACCCCTTAAactcggcctctgtctctccatcttaccactgcccagggtatggttat tgccatctgcacatctgctcttaccatgtctcctactacaacacatcat[g/c]gcctgggcg ctataactctcatctctctcctcagggaccagctgctcctggaccagctgcaagaactcctggaa cactggcaactgcaccaattacttctccgaggacaacatcacctggacctccctccactccacgtccc ctgctgaagaattttacacgcgccacgtaatgcacgtatgaggttggCCTGTTAGGGGCAGG CCA
SLC6A4 u6	SLC6A4	C	T	F	F	cds	GE1124	CCGTGGGCCA CCCTTA	TGGCCTGCCCC TAACAGG	CCGTTGGGCCACCCCTTAAactcggcctctgtctctccatcttaccactgcccagggtatggttat tgccatctgcacatctgctcttaccatgtctcctactacaacacatcatgctcctgggcgctat actactctcatctcctcctcagggaccagctgctcctggaccagctgcaagaactcctggaaact ggcaactgcaccaattactt[c/t]ccgaggacaacatcacctggacctccactccactccacgtccc ctgctgaagaattttacacgcgccacgtaatgcacgtatgaggttggCCTGTTAGGGGCAGG CCA
SLC6A4 u7	SLC6A4	C	A	-	-	noncoding	GE1027	AGCATCAGTAA CCTGCACACTC	ATTTCAGGCC CGACTGGT	AGCATCAGTAACTGCACACTCTctctccctaggctctcctggaggcgaagg[c/a]gaccttgcctt ccctctattgcagaataacaagggtcttagccacagagtgtgctggcaagtggaaagaagaacaa atggtgagcagcagggaagtttaaggACCAGTCGGGCTTGCATAAT
SLC6A4 u8	SLC6A4	A	G	T	A	cds	GE1079	CTTAGACCCCT GATCTTGA	TCAAAGCAAG CAACTCAG	CTTAGACCCCTGTGATCTTGAactgtctcagggcccccttgggttttccccctccagagatgacct ggta[a/g]ccagcgtggtgaactgcatagcagcctctgcttctgggatttgcattctcacagtg ctcgggttacatggctgagatgaggaatgaagatgtgtctgaggtggccaaagacgaggtaggac ctcgggttctctagcaggcccccttgcctcctacTGAGTGTGCTTTGA
TBXAS1 a15	TBXAS1	C	A	L	I	cds	GE282	CCTTCATCTGC AGCCATTT	TGTACAGCAAT TATGGACAGTT A	CCTTCATCTGCAGCCATTTAggggtgactcccgcatgggtgccctaattacaccttctgttacc ttatcagatggttccccctcatcagcaagcctgcgacct[c/a]tctggctcattttaaaccgc tatgggaaatctggggacgcatttgacatccagaggttaaggctgctgcattacagatgagaatc gagtttctgaatcactgctctctgtTAACGTGCTTAATTAATGCTGACAA

FIG. 5

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Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
TBXAS1 a16	TBXAS1	A	G	-	-	noncoding	GE332	CGAGATTGAAA TTTAAGGAAA GAC	CCAGAAACACA AGTGTAACTG A	CGAGATTGAAATTTAAGGAAAGACAAATgctgagatttgggttaacacga a/g cttctc ccttctcagcagccctccatcagatggccctgagttctgcagcctcgaggaaggccctgccc tctggacatggtgattgcagagacgtgaggtaccgcccagcttcaggtgtggtgagccc cctccctgcccaggtcccacacctaccctaccctgcccagcctgcaggtcagggccct cctcgaTCAGTTACCACTTGTGTTCTGG	289
TBXAS1 a17	TBXAS1	C	T	-	-	noncoding	GE332	CGAGATTGAAA TTTAAGGAAA GAC	CCAGAAACACA AGTGTAACTG A	CGAGATTGAAATTTAAGGAAAGACAAATgctgagatttgggttaacacga a/g cttctc tctcagcagccctccat c/t agatggccctgagttctgcagcctcgaggaaggccctgccc tctggacatggtgattgcagagacgtgaggtaccgcccagcttcaggtgtggtgagccc cctccctgcccaggtcccacacctaccctaccctgcccagcctgcaggtcagggccct cctcgaTCAGTTACCACTTGTGTTCTGG	289
TBXAS1 d12	TBXAS1	C	G	Q	E	cds	GE257	TGCTGTTCCAA ATTGTTTACTG A	TTCACACGCTG AAATCAAAAT	TGCTGTTCCAAATTTTACTGAAataagtttgaataatttggaatttctgcttaattcttctac tatagtgctgtgttctctcaggggtttttgggaaagc c/g aaatggagctcagaagctgta tggacctctgtggtggaagaagaactcaaccttctattatgtacgatatcttctctattatgt acgatATTTTGAATTTTCACGCTGTGAA	222
TBXAS1 d13	TBXAS1	G	A	R	H	cds	GE326	TGGAAACCTAT TCTTTTGCCCT T	TACAGCCATGA GCCACTGT	TGGAAACCTATTTTGTGCTTactccagagagctcagtaattctaggttctcctaatagagcct aaagcatgagtgcaacttcatctcagcttctgaaattctgcttctccctccaggtactccacat cagcatctcgaagctggagaagttagcctcagacatcccaagccttctcttctcattggaaac ttgacatttctcc g/a ccaggtgaaggctgtctccattggcttccatcataaaatattgctga gggacagggCACAGTGGCTCATGGCTGTA	288
TBXAS1 d14	TBXAS1	G	A	-	-	noncoding	GE355	CTTGAGCATC CTTGCTCTCA	GCTCTCACGCA GAGAACTGG	CTTGAGCATCTCTGCTCTCAGatgcagggtggctcagctggagacagggctgcagaggggaggg gagcgggtgttctggggccagccctgaccacacaggaacctgcaggttcacggctgagggccggcagc agacccggcccttcacgtacctgctcctcgggccggccacagggctgctcgggtgcatcta gggtgcttgaggtcaagttgacactgctccagctgcacaggtccgggtccaaagctgccc tgagacccaggtgagggccctcctcagagggcag g/a tacaggggcagcgggtgggaggggcca ccccAGTTCTCTGCGTGAGAGC	347
TBXAS1 u1	TBXAS1	G	C	V	L	cds	GE355	CTTGAGCATC CTTGCTCTCA	GCTCTCACGCA GAGAACTGG	CTTGAGCATCTCTGCTCTCAGatgcagggtggctcagctggagacagggctgcagaggggaggg gagcgggtgttctggggccagccctgaccacacaggaacctgcaggttcacggctgagggccggcagc agacccggcccttcacgtacctgctcctcgggccggccacagggctgctcgggtgcatcta gggtgcttgaggtcaagttgacactgctccac g/c tgctgcaaaagtccgggtccaaagcct gacctgagacccaggtgagggccctcctcagagggcaggtacaggggagcgggtgggaggggcca ccccAGTTCTCTGCGTGAGAGC	347
TBXAS1 u10	TBXAS1	A	C	-	-	noncoding	GE249	ATGGACCTGTA TTGCCACCA	GAGAGTTTGA TTTCTCATGTC TTA	ATGGACCTGTAATGGCCACCAaggltgggttctggctccctgagtcctgacctctgctt a/c cttcccaacagcgtcgggttctgaggttcaagtcaggtgagcagcagcttctgcttctacgtgac aaaagatgggagaggtcagaggtgcccctgagctgctgcttctcagtcctgaaaagctgaacgaggt AAGACATGAGAAATGCAACTCTC	219
TBXAS1 u11	TBXAS1	C	T	T	M	cds	GE355	CTTGAGCATC CTTGCTCTCA	GCTCTCACGCA GAGAACTGG	CTTGAGCATCTCTGCTCTCAGatgcagggtggctcagctggagacagggctgcagaggggaggg gagcgggtgttctggggccagccctgaccacacaggaacctgcaggttcacggctgagggccggcagc agcaccggccctca c/t gtacctgacctcggggccggccacagggctgctcgggtgcatcta tctagggctgctgaggtcaagttgacactgctccacgtgctgcaaaagtccgggtccaaagcct ggcctgagacccaggtgagggccctcctcagagggcaggtacaggggagcgggtgggaggggcca ccccAGTTCTCTGCGTGAGAGC	347

FIG. 5JJJJJJ

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
TBXAS1 u2	TBXAS 1	G	A	A	T	cds	GE274	CGACCTGGTGT TTCCCTCA	TGCTGCCCTCCA CTGGTAAAT	CGACCTGGTGT TTCCCTCAGattcacacgggagggcagctcaggaactgcgaggtgctggggcagcg catccccgcaggc [g/a]ctgtgctagagatggccgtgggtgcccctgaccatgacctgagcac tggccaaagcccgagagacctcaaccctgaaaggtagtactgccccctttaaaaagctctgaaag gatgtgagtggtgtgggagatagaaATTACACAGTGGAGGCGACGA	237
TBXAS1 u3	TBXAS 1	C	T	A	A	cds	GE274	CGACCTGGTGT TTCCCTCA	TGCTGCCCTCCA CTGGTAAAT	CGACCTGGTGT TTCCCTCAGattcacacgggagggcagctcaggaactgcgaggtgctggggcagcg catccccgcaggcgtgtgctagagatggc [c/t]gtgggtgcccctgaccatgacctgagcac tggccaaagcccgagagacctcaaccctgaaaggtagtactgccccctttaaaaagctctgaaag gatgtgagtggtgtgggagatagaaATTACACAGTGGAGGCGACGA	237
TBXAS1 u4	TBXAS 1	C	G	Q	E	cds	GE274	CGACCTGGTGT TTCCCTCA	TGCTGCCCTCCA CTGGTAAAT	CGACCTGGTGT TTCCCTCAGattcacacgggagggcagct [c/g]aggactgcgaggtgctggggc agcgcatccccgcaggcgtgtgctagagatggccgtgggtgcccctgaccatgacctgagcac tggccaaagcccgagagacctcaaccctgaaaggtagtactgccccctttaaaaagctctgaaag gatgtgagtggtgtgggagatagaaATTACACAGTGGAGGCGACGA	237
TBXAS1 u5	TBXAS 1	G	A	R	Q	cds	GE355	CTTGGAGCATC CTGTGCTCA	GCTCTCACGCA GAGAACTGG	CTTGGAGCATCCTTCTCAGatgcaggggtggctcagctggagcaacagggctgcagaggggaggg gagcgggtgtcttggggcagccctgaccacacgagacctgcaggttcacggctgagggccggcagc agcacggcccttcacgtacctgcccctcggggcggccacggagctgcctcgggtgcatcta gggtgctgaggtcaagttgacactgtccacgtgtgcacaaagtcc [g/a]gttccaaagcct gccctgagacccaggtgagggccccctgtcagagggcaggtacaggggagcggtggggagggcca cccacagttcttgcgtgagagc	347
TBXAS1 u6	TBXAS 1	T	G	V	G	cds	GE470	GGCCTGGTGT ATTATCACC	CCAAAGTGGC TCCATTC	GGCCTGGTGTATTATCACCccccctttccaatgccactttgtttttctcttccaagtatcttcc catccataatgggtcccctggcccgaattttgcccaataaagaccgagacgaactgaatggcttt ttaacaaactcattaggaatg [t/g]gattgcttgcgggaccagcaagctgccgaagaggttaa cgtatttaataaggacacagccttgaatgGAATGGAGCGACGCTTTCG	243
TBXAS1 u7	TBXAS 1	T	C	I	T	cds	GE912	GCCCATGTATC TTCCCTCCTTT	GGGGATCCAA CTGTACT	GCCCATGTATCTTCCCTCTTgttctccaggaagcctcactcttcatgactgaaggtcaaaatg tgcattttctccttctgttctttagaggcgagagacttctcccaatggctcctggatgccga catctgcaagtcctccatggcggtgcaagactttgacatcgtcagagagcttttctcctctactgg gtgcaagccgaaccttcccggcaacacacagccacccatggccagggcctttgactgtggatg aga [t/c]gtggggcagggccttcatcttctcatcgtggtgctatgaaatcatcaccacacact tcttggccacctactgtgcccacacccctgactgccaagagagcttctgagagaggtag acgtttttaaaggagaaacacgtgAGTACAAGTTGGATCCCC	432
TBXAS1 u8	TBXAS 1	C	G	L	V	cds	GE912	GCCCATGTATC TTCCCTCCTTT	GGGGATCCAA CTGTACT	GCCCATGTATCTTCCCTCTTgttctccaggaagcctcactcttcatgactgaaggtcaaaatg tgcattttctccttctgttctttagaggcgagagacttctcccaatggctcctggatgccga catctgcaagtcctccatggcggtgcaagactttgacatcgtcagagagcttttctcctctactgg gtgcaagccgaaccttcccggcaacacacagccacccatggccagggcctttgactgtggatg agattgtggggcagggccttcatcttctcatcgtggtatgaaatcatcaccacacacttct tttggccacctac [c/g]tactggccacacacccctgactgccaagagagcttctgagagaggtag acgtttttaaaggagaaacacgtgAGTACAAGTTGGATCCCC	432
TBXAS1 u9	TBXAS 1	T	G	M	R	cds	GE274	CGACCTGGTGT TTCCCTCA	TGCTGCCCTCCA CTGGTAAAT	CGACCTGGTGT TTCCCTCAGattcacacgggagggcagctcaggaactgcgaggtgctggggcagcg catccccgcaggcgtgtgctagagat [t/g]ggccgtgggtgcccctgaccatgacctgagcac tggccaaagcccgagagacctcaaccctgaaaggtagtactgccccctttaaaaagctctgaaag gatgtgagtggtgtgggagatagaaATTACACAGTGGAGGCGACGA	237

FIG. 5KKKKKKK

FIG. 5LLLLLL

[illegible]

FIG. 5MMMMMM

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FIG. 5NNNNNNN

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
THPOu4	THPO	A	T	K	M	cds	GE416	GGCATCTCTGTC TTCTCTACTTA GAC	AGGAATCTTG TCCAGTGTCT C	GGCATCTCTGTC TTCTCTACTTA GACaaggaggcctgagatctggccctgggttttgccctcagg accatcctctgcccctcagcttctccacacaggcaggaccacagctcacaaggatcccaatgccat ctctctgagcttccacacacctgctccgaggaa [a/t] ggtgcgttctctgagctgtaggaggg tccacccctctgctcaggcgggcccacacacacacagctgtcccagagaaacctctctagctct cacactgaacgagctcccaaacaggacttctggattgttggagacaaacttccactgacctcagcca gaactactggctctgggctctgaagtgagcagagggaattcagagcagaagattcctggctctgctg aatcaaacctccaggctccctggacaaatcccgggataacctgaacaggatacacgaactcttgaa tggaaactgtggactcttctctggaccctcagcagagaccctaggagccccggacatttctctcag gaactcagacaggctccctggacacccacacacacacacacacacacacacacacacacacacac cctctactggacagataacgtctctctctctccacacacacacacacacacacacacacacacac ccaacccctgcttctgaccttctgctcacaacacacacacacacacacacacacacacacacacac cctac tctgctctgctgacagctcccttccctgacaggcggcccttggGAGACAACTGGACAGATTTCCT	844
THu1	TH	G	A	S	S	cds	GE1125	GGTCCCCGGGT CTCAGC	AGCCCCACCCA CAGGTGA	GGTCCCCGGGTCTCAGCagggtgaggaggcattgggtgccccctgtccccacagtcctccgcggttc atttggggcgcagcagagagcctcatcgaggacgcccgcgaaggagcggaggcgggtggcagcagc ggcgcgtgagctccctcggagcccggggaccccttggagctgtggccttggaggagaaggagg ggaaggcc [g/a] tgcataaacctgctcttctccccgaggggccacacacacacacacacacacac agctgtgaaagggtgttggaggtagctgtggccttctgtccccctgggggcaagtTCACCTGTGGGT GGGGCT	331
THu2	TH	G	A	V	M	cds	GE1125	GGTCCCCGGGT CTCAGC	AGCCCCACCCA CAGGTGA	GGTCCCCGGGTCTCAGCagggtgaggaggcattgggtgccccctgtccccacagtcctccgcggttc atttggggcgcagcagagagcctcatcgaggacgcccgcgaaggagcggaggcgggtggcagcagc ggcgcgtgagctccctcggagcccggggaccccttggagctgtggccttggaggagaaggagg ggaaggcc [g/a] tgcataaacctgctcttctccccgaggggccacacacacacacacacacacac agctgtgaaagggtgttggaggtagctgtggccttctgtccccctgggggcaagtTCACCTGTGGGT GGGGCT	331
THu3	TH	T	G	F	C	cds	GE1125	GGTCCCCGGGT CTCAGC	AGCCCCACCCA CAGGTGA	GGTCCCCGGGTCTCAGCagggtgaggaggcattgggtgccccctgtccccacagtcctccgcggttc atttggggcgcagcagagagcctcatcgaggacgcccgcgaaggagcggaggcgggtggcagcagc ggcgcgtgagctccctcggagcccggggaccccttggagctgtggccttggaggagaaggagg ggaaggccgtgtataaacctgctcttctccccgaggggccacacacacacacacacacacacacacac gtgaagggtgt [t/g] tgaaggtagctgtggccttctgtccccctgggggcaagtTCACCTGTGGGT GGGGCT	331
THu4	TH	G	A	K	K	cds	GE1020	CTGCCCCGAGG AAGGAG	CTGGGCACACC CTTCAG	CTGCCCCGAGGAGGAGgtctacaccacgctgaa [g/a] ggcctctacgccacacgacctgagg ggaacacctggaggccttctgttggagcgttctcagcggctacccgggaagacaataatcccccc agctggaggagagctctcccgttctCTGAAGGGGTGTGCCAG	170
THu5	TH	T	C	A	A	cds	GE1125	GGTCCCCGGGT CTCAGC	AGCCCCACCCA CAGGTGA	GGTCCCCGGGTCTCAGCagggtgaggaggcattgggtgccccctgtccccacagtcctccgcggttc atttggggcgcagcagagagcctcatcgaggacgcccgcgaaggagcggaggcgggtggcagcagc ggcgcgtgagctccctcggagcccggggaccccttggaggt [t/c] gtggccttggaggagaag gaggggagggcgtgtataaacctgctcttctccccgaggggccacacacacacacacacacacacacacac agctgtgaaagggtgttggaggtagctgtggccttctgtccccctgggggcaagtTCACCTGTGGGT GGGGCT	331
THu6	TH	G	T	A	A	cds	GE972	TACGCGCAGGG ACTGCT	GGGTACCTGGG AGAACT	TACGCGCAGGACTGCTgccacgagctgtgggggacgtgccccatgctggccgacccgacacctcg c [g/t] cAGTTCTCGCAGGTAGC	89

FIG. 50000000

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
VLDLRd 13	VLDLR	C	T	A	A	cds	GE926	GAAAGACCTTG CCTTCTTAAAG C	CAAGTGACAAT GACTTATGTCA AGA	GAAAGACCTTGCTTCTTAAAGCaaactaagtaacccagacttccatcttgaggcaaaagagcc aagtctgacttcaactaactcgaagagacatcaggaagattggcttagagagagaaagaaatataccc aactagttgaacagactaagaaacacttggtctcgatgctgacattgctgcccagaactatctc tgggc(c/t)gactaagccaaaggctatcttcaggtaacttccagttccttttgggtgTCTT GACATAAGTCAATTGTCACTTG	281
VLDLRd 14	VLDLR	A	T	-	-	noncoding	GE926	GAAAGACCTTG CCTTCTTAAAG C	CAAGTGACAAT GACTTATGTCA AGA	GAAAGACCTTGCTTCTTAAAGCaaactaagtaaccc(a/t)gacttccatcttgaggcaaaag agccaagtctgacttcaactaactcgaagagacatcaggaagattggcttagagagagaaagaaatata atccaactagttgaacagctaaagaaacacttggtctcgatgctgacattgctgcccagaact attctgggcccgatctaaagccaaaggctatcttcaggtaacttccagttccttttgggtgTCTT GACATAAGTCAATTGTCACTTG	281
VLDLRd 15	VLDLR	G	T	-	-	noncoding	GE937	AGGTTTGGCT CCTTACC	GGTAGCTCCAG ATGAACAAAA	AGGTTTGGCTCCTTACCTgatgggtaaatttctaaagctgaatacacagatccttctaaactgatt cctttattcctctgtaggagatcaatgagacacagagatcagaggtcagtggttccccaaaa gggacttctgcccagatgggccaattcttctctctctgtaagta(g/t)atttctctannngtctgggt tcaagaaacttcttagataccagatgaagatTTTTTGTTCATCTGGAGCTACC	247
VLDLRd 16	VLDLR	G	A	E	K	cds	GE940	CCTGGTGT AATGTGAAAGA TA	TATCCTTTCCC ATCACCTGC	CCTGGTGTAAATGTGAAAGATATtaattgaaaaaattggtcaagtgantantacatttttat tccagatataaacgaatgcttggttaataatggtggaattctctcatatctgcaagacactagta taggctac(g/a)agtgactgactgagctgggttgaactgataagtaaggaaacactgctggaggg tgagtcataagaaagaaacclggacctGCAGGTGATGGAAAGGATA	242
VLDLRd 17	VLDLR	C	T	N	N	cds	GE953	TTTTCACAGCT TTGTTTACTGG T	TGAAGATAGTT GAGTGGGTGGT	TTTTCACAGCTTTGTTTACTGTGtagctgggtgaacagctaaaatagaaagacaggaatga atggattcgatagacgtccactgggtgacagcggaatccagctggcctaa(c/t)ggaattacact tggtatgtatgtctctctctcgcACCAACCCACTCAACTATCTTCA	176
VLDLRu 1	VLDLR	A	G	T	A	cds	GE920	GCTCTAAATTGT GTCAAACTCTT AAAT	GACCTACACAG ATACCATTCCA AAG	GCTCTAAATTGTGTCAAACTCTTAAATtcttgtagactattctgttccagtgccctcaattgatga caaggttggttagacatgttaaaatgacgacaaatgctataatcctgcagccattgctgttgatt gggtgacaaagacactctactggactgactgagcttcttaagactatttcaagtagctacctagat gga(a/g)ccaagaggaagtctctgtttaactctgacttgagagagcctgcccataagctgtgg acceaactgtctgggtttagtctgttctccatcacagaCTTTGGAAATGTATCTGTGTAGGTC	324
VLDLRu 10	VLDLR	A	T	S	C	cds	GE937	AGGTTTGGCT CCTTACC	GGTAGCTCCAG ATGAACAAAA	AGGTTTGGCTCCTTACCTgatgggtaaatttctaaagctgaatacacagatccttctaaactgatt cctttattcctctgtaggagatcaatgagacacagcagatcagaggtc(a/t)tggttcccc aaaagggaacttctgcccagatgggccaattcttctctgtaagtagatttctctannngtctgggt tcaagaaacttcttagataccagatgaagatTTTTTGTTCATCTGGAGCTACC	247
VLDLRu 11	VLDLR	T	C	I	I	cds	GE937	AGGTTTGGCT CCTTACC	GGTAGCTCCAG ATGAACAAAA	AGGTTTGGCTCCTTACCTgatgggtaaatttctaaagctgaatacacagatccttctaaactgatt cctttattcctctgtaggagatcaatgagacacagcagatcagaggtcagtggttccccaaaa gggacttctgcccagatgggccaattcttctctgtaagtagatttctctannngtctgggt tcaagaaacttcttagataccagatgaagatTTTTTGTTCATCTGGAGCTACC	247
VLDLRu 12	VLDLR	A	T	S	C	cds	GE945	TCCAATACTAG ACTTAGCTCAC TT	GACTTACTGCT GGGTACCTG	TCCAATACTAGACTTAGCTTACTagctaccctctgatttttctcagtgctcttagtgatggcag cagtaggtggctacttgatgtggcggaattggcaacacagaacatgaaagcatgaactttgac aatcctgtgtacttgaaacacactgaagaggaacctctccatagacattggtagacac(a/t)gtg cttctgttggaacACGTACCCAGCAGTAAGTC	228
VLDLRu 2	VLDLR	A	G	K	R	cds	GE926	GAAAGACCTTG CCTTCTTAAAG C	CAAGTGACAAT GACTTATGTCA AGA	GAAAGACCTTGCTTCTTAAAGCaaactaagtaacccagacttccatcttgaggca(a/g)ag agccaagtctgacttcaactaactcgaagagacatcaggaagattggcttagagagagaaagaaatata atccaactagttgaacagctaaagaaacacttggtctcgatgctgacattgctgcccagaact attctgggcccgatctaaagccaaaggctatcttcaggtaacttccagttccttttgggtgTCTT GACATAAGTCAATTGTCACTTG	281

FIG. 5PPPPPPP

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
VLDLRu 3	VLDLR	A	G	R	R	cds	GE920	GCTCTAATGTG GTCAAACTCTT AAAT	GACCTACACAG ATACCAATTCCA AAG	GCCTTAATTTGCTCAAACTCTTAAATTTCTTGTGACCTATCTGTTCAGTGCCTCAATGATGAGAGGTTGGTGTACAAAGACCATCTACTGGAGTGTGCGGTCTCTAAGACTATTTCAAGTAGTACCTAGATGGGACCAAGAGTCTCTGTTAACTCTTGGAAATGGTATCTGTGTAGGTC	324
VLDLRu 4	VLDLR	A	G	Q	Q	cds	GE934	GAATGGACTT GTGTTAATCCT G	TGCTGCTTCCC TTAAAGTAGTT AT	GAATGGACTTGTGTAAATCTGTGATGTACATGCTAATTTGTGGGCTCTGTCTTAGGATCGTGTCTTACTGGATAGTGGGAAATGAAGCAGTCTGTGGTGCCTAATAATTCAGTGCAGAGCTAGCCCTAGTCAACAACCTGATGCCA[la/g]GACATCATCTCTATCATGAAGTGTACAGCCATCAGGTACCGTGGAGAACAGTCTCTAATAACTACTTTTAAGGGAGAGCA	251
VLDLRu 5	VLDLR	A	C	E	A	cds	GE941	GCACCGAATA CCCATTTTA	TGCTTGTGTGT AGGTTCTACTT GTG	GCACCGAATAACCCATTTTAATGATGATTTCTTCTGACTAGTAAATAATTTGGTGTGAGAGAGACATGG[la/c]GAAATGGAGATGTGAATACCTATGCTGCCAGCACACAGATTAATGATCACTCTCCAAATATACCTGTCTGCTCCAGTGGTACAAATGAGAGGAAATGGCGAGAGCTGTCAAAGTAAGGCATTTTGTGTTCAACACAAAGTAGAACCTACACAAAGCAA	241
VLDLRu 6	VLDLR	C	T	L	F	cds	GE944	TGTACCTAGTA AGGTATAGGAG CAGC	CACCTACTCAT AAAGGTACAAAC AGCC	TGTACCTAGTAAAGTATAGGAGCAGCAAGACTAATCTGATTTCTCTCCAGATATTGATGAATGCCAAATCCAGGAATCTGCAATCTGATCAACTAAAGGCGGTACAAGTGTGAATGTGCTGGCTATCAAAATGGAT[cl]TTGCTACTGGGCTGTGCAAGGCAAGTAAATGAAGTGGTGTATGCTGTCTTATGAGTAAGTG	230
VLDLRu 7	VLDLR	G	A	V	I	cds	GE236	ATTCTAGGGAG AAAGCCAAA	TTTACTTACCA CAGTTCTTTTC ATCA	ATTCTAGGGAGAAAGCCAAAATGTGAACCTCTCCCAATCTCCAGTGCACAAATGGTCTGCTATTACGCTGTGTGGAATGTGATGGGATGAAGACTGT[ga]TTGACGGCAGTCATGAAAGCAACTGTGGTAAGTAA	140
VLDLRu 8	VLDLR	G	C	C	S	cds	GE911	CCAACTTTGAT GCATTTTTCAG	CAGTTGATACA GGGAAAGAAC TG	CCAACTTTGATGCAATTTTCAGTGGGCACTCTCTCTCTAATAGGCAATAAATGATGCTCCGACGATTCACCTGTCCAGTGGCGCTGCATCTCCAGAACCTTGTATGCAATGGCCAGGATGACGCAAGCAGTGGCAGTGTGAGTGGAGTGTGCCCCCACTGTGGCGCCATGAGTCCAGT[ga/c]CAGCAGCTCTCTGCTGCATCCCCATCAGCTGGGTATGCAAGATGTGCAAGACTGCTCCGACCAATCTGATGAGTCCCTGGAGCAGTGTGGCGCTCAGCAGTGCATCACACACCAAGTGTCCAGCCAGCGAAATCCAGTGGGCTCTGGGAGTGTGATCCATGAAGTGGGAGTGTGATGGGAGCCCTGACTGCAAGGATGGCAGTGTGAGGTCAACTGTCTGTAAGTGTCTCTAGCATGGCATGTTCCCTGTATCAACTG	471
VLDLRu 9	VLDLR	T	A	I	N	cds	GE941	GCACCGAATA CCCATTTTA	TGCTTGTGTGT AGGTTCTACTT GTG	GCACCGAATAACCCATTTTAATGATGATTTCTCTCTGACTAGGTAATAATTTGGTGTGAAGAGACATGGAGATGGAGATGTGAATACCTATGCTGCCAGCACCAAGA[t/a]TAATGATCACTCTCCAAATATACCTGTCTGCTCCAGTGGGTACAAATGAGAGGAAATGGCGAGAGCTGTCAAAGTAAGGCATTTTGTGTTCAACACAAAGTAGAACCTACACAAAGCAA	241

FIG. 5QQQQQQQ

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(74) Agents: **HOGLE, Doreen, M. et al.**; Hamilton, Brook, Smith & Reynolds, P.C., Two Militia Drive, Lexington, MA 02421 (US).

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(84) Designated States (*regional*): ARIPO patent (GH, GM, KE, LS, MW, SD, SL, SZ, TZ, UG, ZW), Eurasian patent (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM), European patent (AT, BE, CH, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE), OAPI patent (BF, BJ, CF, CG, CI, CM, GA, GN, GW, ML, MR, NE, SN, TD, TG).

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23 August 2001

For two-letter codes and other abbreviations, refer to the "Guidance Notes on Codes and Abbreviations" appearing at the beginning of each regular issue of the PCT Gazette.

WO 00/58519 A3

(54) Title: CHARATERIZATION OF SINGLE NUCLEOTIDE POLYMORPHISMS IN CODING REGIONS OF HUMAN GENES

(57) Abstract: The invention provides nucleic acid segments of the human genome, particularly nucleic acid segments from the coding region of a gene, including polymorphic sites. Allele-specific primers and probes hybridizing to regions flanking or containing these sites are also provided. The nucleic acids, primers and probes are used in applications such as phenotype correlations, forensics, paternity testing, medicine and genetic analysis.

INTERNATIONAL SEARCH REPORT

International Application No
PCT/US 00/08440

A. CLASSIFICATION OF SUBJECT MATTER

IPC 7 C12Q1/68

According to International Patent Classification (IPC) or to both national classification and IPC

B. FIELDS SEARCHED

Minimum documentation searched (classification system followed by classification symbols)

IPC 7 C12Q

Documentation searched other than minimum documentation to the extent that such documents are included in the fields searched

Electronic data base consulted during the international search (name of data base and, where practical, search terms used)

C. DOCUMENTS CONSIDERED TO BE RELEVANT

Category *	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
X	WO 96 20279 A (RIBOZYME PHARM INC ;WARNER LAMBERT CO (US)) 4 July 1996 (1996-07-04) p. 15, sequence "013a"	1
X	US 5 705 388 A (COUTURE L. ET AL.) 6 January 1998 (1998-01-06) SEQ ID 622,623	1

☒ Further documents are listed in the continuation of box C.

☒ Patent family members are listed in annex.

* Special categories of cited documents :

- "A" document defining the general state of the art which is not considered to be of particular relevance
- "E" earlier document but published on or after the international filing date
- "L" document which may throw doubts on priority claim(s) or which is cited to establish the publication date of another citation or other special reason (as specified)
- "O" document referring to an oral disclosure, use, exhibition or other means
- "P" document published prior to the international filing date but later than the priority date claimed

- "T" later document published after the international filing date or priority date and not in conflict with the application but cited to understand the principle or theory underlying the invention
- "X" document of particular relevance; the claimed invention cannot be considered novel or cannot be considered to involve an inventive step when the document is taken alone
- "Y" document of particular relevance; the claimed invention cannot be considered to involve an inventive step when the document is combined with one or more other such documents, such combination being obvious to a person skilled in the art.
- "&" document member of the same patent family

Date of the actual completion of the international search

10 November 2000

Date of mailing of the international search report

22.02.01

Name and mailing address of the ISA

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Authorized officer

Luzzatto, E

INTERNATIONAL SEARCH REPORT

Interr 1al Application No
PCT/US 00/08440

C.(Continuation) DOCUMENTS CONSIDERED TO BE RELEVANT		
Category *	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
A	<p>DATABASE MEDLINE [Online] American medical association Accession Number 92172843, 3 March 1992 (1992-03-03) XP002152537 abstract & SUMI-ICHINOSE C. ET AL.: "Molecular cloning of genomic DNA and chromosomal assignment of the gene for human aromatic L-amino acid decarboxylase, the enzyme for catecholamine and serotonin biosynthesis" BIOCHEMISTRY, vol. 31, no. 8, 1992, pages 2229-2238, US</p>	1,11
A	<p style="text-align: center;">---</p> <p>CHEE M ET AL: "ACCESSING GENETIC INFORMATION WITH HIGH-DENSITY DNA ARRAYS" SCIENCE,US,AMERICAN ASSOCIATION FOR THE ADVANCEMENT OF SCIENCE,, vol. 274, 25 October 1996 (1996-10-25), pages 610-614, XP002022508 ISSN: 0036-8075 cited in the application the whole document</p>	1-12
A	<p style="text-align: center;">---</p> <p>WANG D G ET AL: "Large-scale identification, mapping, and genotyping of single-nucleotide polymorphisms in the human genome" SCIENCE,US,AMERICAN ASSOCIATION FOR THE ADVANCEMENT OF SCIENCE,, vol. 280, 1998, pages 1077-1082, XP002089398 ISSN: 0036-8075 cited in the application the whole document</p>	1-12
A	<p style="text-align: center;">---</p> <p>OEFNER P J ET AL: "COMPARATIVE DNA SEQUENCING BY DENATURING HIGH-PERFORMANCE LIQUID CHROMATOGRAPHY" AMERICAN JOURNAL OF HUMAN GENETICS,UNIVERSITY OF CHICAGO PRESS, CHICAGO,,US, vol. 57, October 1995 (1995-10), page A266 XP002918646 ISSN: 0002-9297 cited in the application the whole document</p>	1-12
A	<p style="text-align: center;">---</p> <p>WO 98 20165 A (WHITEHEAD BIOMEDICAL INST ;HUDSON THOMAS (US); LANDER ERIC S (US);) 14 May 1998 (1998-05-14) page 4, line 1 -page 27, line 23 ---</p> <p style="text-align: center;">-/--</p>	1-12

INTERNATIONAL SEARCH REPORT

Internal Application No
PCT/US 00/08440

C.(Continuation) DOCUMENTS CONSIDERED TO BE RELEVANT		
Category *	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
A	<p>DATABASE GENE BANK [Online] accession number M84597, 18 April 1992 (1992-04-18) ICHINOSE H. ET AL.: "Molecular cloning of genomic DNA and chromosomal assignment of the gene for human aromatic L-amino acid decarboxylase" XP002152538 abstract</p>	
A	<p>--- SYVANEN A -CH ET AL: "IDENTIFICATION OF INDIVIDUALS BY ANALYSIS OF BIALLELIC DNA MARKERS, USING PCR AND SOLID-PHASE MINISEQUENCING" AMERICAN JOURNAL OF HUMAN GENETICS, US, UNIVERSITY OF CHICAGO PRESS, CHICAGO,, vol. 52, no. 1, 1993, pages 46-59, XP002050638 ISSN: 0002-9297 the whole document</p>	11,12
A	<p>--- UNDERHILL P.A. ET AL.: "Detection of numerous Y chromosome biallelic polymorphisms by denaturing high-performance liquid chromatography" GENOME RESEARCH, vol. 7, - 1997 pages 996-1005, XP000942730 US abstract</p> <p>-----</p>	1-12

INTERNATIONAL SEARCH REPORT

In. ational application No.
PCT/US 00/08440

Box I Observations where certain claims were found unsearchable (Continuation of Item 1 of first sheet)

This International Search Report has not been established in respect of certain claims under Article 17(2)(a) for the following reasons:

1. ☐ Claims Nos.:
because they relate to subject matter not required to be searched by this Authority, namely:
2. ☐ Claims Nos.:
because they relate to parts of the International Application that do not comply with the prescribed requirements to such an extent that no meaningful International Search can be carried out, specifically:
3. ☐ Claims Nos.:
because they are dependent claims and are not drafted in accordance with the second and third sentences of Rule 6.4(a).

Box II Observations where unity of invention is lacking (Continuation of Item 2 of first sheet)

This International Searching Authority found multiple inventions in this international application, as follows:

1. ☐ As all required additional search fees were timely paid by the applicant, this International Search Report covers all searchable claims.
2. ☐ As all searchable claims could be searched without effort justifying an additional fee, this Authority did not invite payment of any additional fee.
3. ☐ As only some of the required additional search fees were timely paid by the applicant, this International Search Report covers only those claims for which fees were paid, specifically claims Nos.:
4. ☒ No required additional search fees were timely paid by the applicant. Consequently, this International Search Report is restricted to the invention first mentioned in the claims; it is covered by claims Nos.:

1-12 all partly

Remark on Protest

- ☐ The additional search fees were accompanied by the applicant's protest.
- ☐ No protest accompanied the payment of additional search fees.

FURTHER INFORMATION CONTINUED FROM PCT/ISA/ 210

1. Claims: 1-12 (all partly)

Nucleic acid molecules comprising the polymorphic sites present in the sequences SEQ ID 1927 and 1928 given on fig. 5A (which correspond to the sequences designated AADCd4 and AADCd5 in fig. 5A), and methods based thereon.

2. Claims: 1-12 (all partly)

Nucleic acid molecules comprising the polymorphic site(s) present in each of the sequences given on figs. 5A (except the sequences AADCd4 and AADCd5), 5B-5QQQQQQ and methods based thereon.

The sequences differing only at one or more polymorphic sites are considered to belong to the same invention. The different inventions are summarised as indicated in the following table (see annex), where col. 1 and 3 show the SEQ ID number and col. 2 and 4 the length of the sequence. The total number of inventions is 396.

INTERNATIONAL SEARCH REPORT

Information on patent family members

International Application No

PCT/US 00/08440

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